THE MPS MAGAZINE

Society for Mucopolysaccharide Diseases

Support Research Awareness

Summer 2015

www.mpssociety.org.uk

Vimizim Campaign

Catch up with the latest developments in the fight for funding for Vimizim.

Page 5

MPS Weekend Conference

Friday 26th June - Sunday 28th June saw us holding our 25th MPS Conference, bringing together both professionals and families to share information and personal stories.

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MPS Awareness Day 2015

The 15th May saw our supporters Wearing Blue in support of MPS Awareness Day.

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Front cover photo: Jibreel Arshad

Society for Mucopolysaccharide Diseases

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Registered as a Company limited by guarantee in:
England & Wales No. 7726882
Registered Charity No. 1143472
Charity registered in Scotland SCO41012

Magazine Deadlines

Autumn	1 September 2015
Winter	1 December 2015
Spring	1 March 2016
Summer	1 June 2016

To submit content email;
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The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee.

The MPS Society reserves the right to edit content as necessary. Products advertised in this magazine are not necessarily endorsed by the Society.

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New Awareness Poster

For those of you looking to raise awareness for MPS and related diseases, you may be interested to learn that we have a brand new A3 poster available.

The poster will be included in our fundraising packs or you can request these separately by emailing fundraising@mpssociety.org.uk. You can also download a printable version from our website by looking under the 'Fundraising Downloads' section.

The MPS Society

Founded in 1982, the Society for Mucopolysaccharide Diseases (the MPS Society) is the only national charity specialising in MPS and Related Diseases in the UK, representing and supporting affected children and adults, their families, carers and professionals.

Our Aims:

To act as a support network for those affected by MPS and Related Diseases.
To promote and support research into MPS and Related Diseases.
To bring about more public awareness of MPS and Related Diseases

MPS and Related Diseases

Mucopolysaccharide (MPS) and Related Diseases affect 1:25,000 live births in the United Kingdom. One baby born every eight days in the UK is diagnosed with an MPS or related disease.

These multi-organ storage diseases cause progressive physical disability and in many cases, neurological deterioration can result in death in childhood.

At present there is no cure for these devastating diseases, only treatment for the symptoms as they arise.

PLEASE NOTE

Our office telephone number has now been changed to 0345 389 9901.

This change will make it more economical for our members and supporters to contact us.

Thank you.

give with confidence

Contents

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twitter.com/MPSSocietyUK

Welcome

We have been very busy since the publication of our last magazine - not only have we celebrated MPS Awareness Day but we also held our biennial MPS Weekend Conference on 26th June to 28th June.

MPS Awareness Day, as always, was a great success, with lots of our families getting involved with schools and workplaces.We have featured a number of your fantastic Wear It Blue stories starting on page 34.

It was lovely to see so many families and professionals over our Weekend Conference - both catching up with familiar faces and meeting new ones. The children also had a great time, much to the credit of our wonderful volunteers, who made it possible for them all to enjoy the weekend's outings and parties. Read some of the highlights on pages 18 - 21.

Lastly you can catch up with the latest Vimizim Campaign news on pages 5 - 7

Best wishes

The MPS Team

Visit our online shop

www.mpssociety.org.uk.

Purchase our information resources and MPS merchandise including our T-shirt!

Chief Executive's Report

Christine Lavery

I am sitting down to write to you reeling from the news that the NICE, the National, Institute of Health and Care Excellence, have indicated that they are leaning towards **NOT** recommending NHS England reimburse Enzyme Replacement Therapy (ERT) Elosulfase Alfa for the treatment of Morquio disease (MPSIVA) in England. When new therapies are trialled clinically the patients and their carers always have to be prepared that the drug may not show benefit but in the case of Morquio disease there is clear evidence that Elosulfase Alfa proved beneficial in stabilising the disease and in the case of young children they continued to grow.

For the 35 children and adults who have been on the clinical trial, and the 42 waiting in the wings steadily deteriorating, and waiting in anticipation that the day was nigh when they will be on commercial Elosulfase Alfa funded by NHS England, this news is utterly devastating. What is more gutting is the knowledge that the NICE process to date has failed these affected children and adults, and their families.

In the MPS Society we have to accept that clinical trials, despite the optimism may not in the future deliver the outcomes we hope for and that new therapies may not prove beneficial to the treatment population. **However in respect of Elosulfase Alfa we believe that this NO decision is the direct result of a failure of due process by NICE NOT appointing adequate and appropriate clinical expertise to provide the vital evidence** . We were so sure of this that on the 8 April 2015 we wrote to the Chair of the Evaluation Committee, Dr Peter Jackson, setting out our considerable concerns. On the 11 May 2015 we received the most breathtakingly patronising response from Meindert Boysen, Programme Director Technology Appraisals of NICE.

Needless to say we are not giving up. Indeed we are determined that not one of our MPSIVA young people who is or could benefit from Elosulfase Alfa will NOT be assisted to deteriorate and die an early death by the arrogant failings of NICE and of NHS England and the Secretary of State for Health, Jeremy Hunt, sleep walking into these failings.

There is now an opportunity for the whole MPS Community of patients, parents, carers and expert clinicians to help make a huge noise politically on behalf of our MPSIVA children and adults to help achieve reimbursed ERT. As an MPS Society we should never lose sight of the fact it is no one's fault that parents bring children into the world who will be diagnosed with MPS, Fabry and related diseases. What is unacceptable is those affected being condemned to a life of unbearable suffering and early death because they have an ultra-rare disease and at the mercy of a Government who prefers to give £1.2 billion to corrupt governments and agencies through the auspices of the International Development Fund. A cut of less than 1% could pay for not just our Morquio members to get ERT but would also pay for boys with Muscular Dystrophy and children and adults dying from tumours associated with Tuberous Sclerosis, to receive their new therapies.

Today the battle is ERT for Morquio but in the near future it will be new therapies for MPSI Hurler, MPSII Hunter, MPSIII Sanfilippo, MPSVI Maroteaux Lamy, MPSVII Sly, Mannosidosis, LaL, Fabry and other lysosomal storage diseases so we need to act now! Please lobby your Member of Parliament and keep lobbying.

I would dearly hope to be sharing better news with you in the next MPS Magazine.

Christine Lavery MBE
Group Chief Executive

Vimizim Campaign

A Year of Campaigning

We had hoped by the time you received this edition of the newsletter we would be able to report that NHS England had finally made a decision on whether to provide interim funding until NICE make their final guidance announcement in October.

However, after a series of false promises on dates NHS England finally sent a press release late on 2nd July to tell us that they have decided that they won't make a decision until October 2015.

The following is a timeline of key dates since Vimizim was licensed by the European Medicines Agency and to date it is now reimbursed to patients in Turkey, Greece, Hungary, France and Germany to name just some of the 30 countries who have been able swiftly decide that treatment will be made available to their citizens with Morquio.

Due to devolved nation healthcare systems we are also working for a decision in Scotland, Wales and Northern Ireland separately – although Wales and Northern Ireland look to NICE's decision rather than having their own process!

Vimizim Campaign Timeline

28th April 2014
On the 28th April 2014 after 5 years of research the drug Elosulfase Alfa, was licensed as Vimizim by the European Medicines Agency, the only treatment currently available for MPSIVA Morquio. The UK had 30% of the patient population enrolled on the clinical trial.

The pharmaceutical company BioMarin continue to provide the treatment compassionately by giving free drug to all 34 patients enrolled on the clinical trial after the drug was licensed by the EMA in April 2014.

21 November 2014
Ten year old Kamal Hoteit sends a letter before proceedings alleging the NHS England scorecard was unlawful and discriminatory to people with ultra-rare diseases. The scorecard had not been published and there had not been any public consultation over its use.

25th November 2014
Greg Mulholland MP raises the uncertainty about funding for a family in his constituency who live in a state of anxiety because they do not know whether the drug Vimizim will be approved for further use.

28 November 2014
NHS England, through Kamal Hoteit's legal team responds, agreeing that the scorecard was unlawful and deferring the decision making process.

2nd December 2014
NHS England reveals it has scrapped key meetings on whether to fund a series of treatments for serious conditions following the threat of legal action.

3rd December 2014
Greg Mulholland MP raises the legal challenge to David Cameron in Parliament and the lack of process.

9th December 2014
On the 9th December 2014 the Minister George Freeman meets with Greg Mulholland MP, three families of children with Morquio and Duchenne Muscular Dystrophy and the two patient organisations, the Society for Mucopolysaccharide Diseases (MPS Society) and Muscular Dystrophy Campaign (MDC).

15th December 2014
A CPAG (Clinical Priorities Advisory Group) decision on reimbursement was expected 15th December but following the legal challenge NHS England scrapped their scorecard system and postponed their decision making.

18th December 2014
On the 18th December 2014 the Minister George Freeman meets with Greg Mulholland MP, the two pharmaceutical companies BioMarin and PTC, and the two patient organisations MPS Society and MDC. George Freeman asks the pharmaceutical companies to submit proposals on treatment cost and delivery. To date we understand that neither of these proposals have been acknowledged.

20th January 2015
Greg Mulholland leads a debate in Westminster Hall about drugs for ultra-rare Diseases.

BioMarin and the MPS Society meet with James Palmer, Richard Jeavons and Edmund Jessop at a stakeholder surgery where it was firmly explained that there would be no interim funding whilst NHS England continued to try to develop a fit-for purpose process. James Palmer stated that the process would include a 90 day stakeholder consultation followed by a CPAG meeting after the General Election and then a 90 day public consultation. This in reality takes us to mid-August 2015 and runs contrary to what the Prime Minister said in PMQ on 4th February 2015.

23rd January 2015
BioMarin send a letter to James Palmer and Richard Jeavons requesting further discussions on their financial offer and suggesting free drug will be stopped if no agreement reached by end of February 2015.

27th January 2015
On the 27th January 2015, NHS England launches a 90 day consultation on their reimbursement decision making process. Once the consultation is complete it will take a CPAG meeting followed by a further 90 days for a decision be made.

4th February 2015
In PMQ David Cameron responds to a question by Greg Mulholland MP about interim funding of Vimizim for Morquio saying that, 'The consultation is under way and will finish at the end of April. Following this, the NHS will make a decision as quickly as possible whether or not to routinely fund Translarna. I have discussed this with the Health Secretary and we will do everything we can to help.'

5th February 2015
On ITV Calendar, when questioned about access for to Vimizim for Sam Brown, David Cameron quotes April for a decision and that the drug company will fund it until May so there is room to make a decision in time.

At the Specialised Health Care Alliance James Palmer suggested that CPAG will sit in May/June and we can expect a decision two weeks after that.

13th February 2015
NICE inform us that the last publication date for NICE guidance consultations on their website will be the 14th March. This is to avoid the release of guidance consultations close to the start of the 'purdah' period. Any guidance or guidance consultations that would have been published in April will now be published in the week beginning 11 May, at the earliest. The guidance planned for publication in May will be issued as normal on Wednesday 27 May.

The Evaluation Committee Meeting for Elosulfase Alfa for treating mucopolysaccharidosis type IVA, which was due to be held on 19 May 2015, has been rescheduled and will now take place on 21 July 2015.

25th March 2015
Thanks to Greg Mulholland's persistence he secures a third meeting with the Minister George Freeman, which was attended by two pharmaceutical company representatives, a number of Duchenne Muscular Dystrophy Charities, Tuberous Sclerosis Association and ourselves. A ten page letter was written to George Freeman highlighting the catalogue of errors from NHS England and how they are failing individuals who are desperate for treatment.

Protests

We are now expert protesters having stood outside the House of Commons with placards and speaking to the press on a number of occassions. Our biggest protest was on April Fool's Day with around 50 of us outside Downing Street with phenomenal placards. We only had to persuade 10 policeman that we weren't going to move and managed to distribute over 500 leaflets.

The protest received a lot of media coverage and Jeremy Hunt said on camera that he would follow this up if he was re-elected. As he is back as Secretary of Health we are following up on his statement.

27th April 2015
The 90 day consultation on process formally ends. Concerns were raised as it lacked detail and methodology.

28th April 2015
One year Anniversary of the EMA licensing Vimizim and the wait for treatment began.

Katy Brown manages to get a one-to-one meeting with David Cameron

May
We utilise the Freedom of Information Act and request information from NHS England on where the money went from PPRS.

We know, according to the Association of British Pharmaceutical Industries, that the pharmaceutical pricing regulation scheme – known as the PPRS – is supposed to be channelling significant sums of money to fund the drugs budget.

This money comes directly from the pharmaceutical industry – as per their agreement with Government at a time of austerity to underwrite the medicines bill between 2014 and 2018. We know that the Department of Health and NHS England through our FOI enquiry cannot account for a staggering £786m - the PPRS rebate in 2014!

It is estimated that the government will receive in excess of £800m from the PPRS this year alone. Sadly, not a penny has gone or will go to the drugs budget in 2015 let alone to fund drugs for ultra-rare diseases like Morquio disease.

12th May 2015
Due to the lack of engagement from NHS England BioMarin had given notice that they would only supply free drug up until this date. However, at the eleventh hour NHS England provides timescales of when a decision will be made which enable BioMarin to continue funding treatment until the 25th June.

Those who were not on the clinical trial are still being denied treatment until a decision is made on the 25th June.

16th June 2015
NICE's committee confirm that they would currently not recommend Vimizim for treating MPSIVA and would like further explanation of the benefits it provides to people with the condition.

Greg Mulholland MP hosted his second parliamentary debate in the House of Commons. The transcript is available on our website. In the debate George Freeman puts forward that the NHS England decision will now be made on 30th June.

NHS England press office however confirm that a decision will be made 1st July 2015

1st July 2015
No decision from NHS England and instead confirm that a decision will be made 2nd July 2015 and that stakeholders will be told under embargo

2nd July 2015
No statement is made until 4:45pm when a press release states that they will not now be making a decision until October 2015.

6th July 2015
MPS Society host a Parliamentary Event with BioMarin (see below), invited MPs, pharmaceutical companies, patient organisations and clinicians to discuss Patient Access to Medicines for Ultra-rare diseases and how this essential but hard fight for Vimizim, and Translarna for Duchenne Muscular Dystrophy can never be repeated.

We have liaised directly with over 45 families with Morquio and their friends and a number of our other MPS families as well as working closely with other patient organisations to join up the dots of the mismatch and discrepancies of information provided by governmental officials. We are then informing our legal team to make sure we do not miss any opportunity to legally challenge when possible.

This campaign has taught us that persistence is key: keep writing to MPs, Ministers, journalists and we will continue to do all we can for a positive NICE decision

Please follow us on Facebook and #fight4treatment and #fundourdrugsNOW on twitter for all the news on the campaign

Charlotte Roberts
Communication Officer

Parliamentary Event for Access to Medicines for Ultra-Rare Diseases

On Monday 6th July we hosted a Parliamentary Event with BioMarin and invited MPs, a number of pharmaceutical company representatives, clinicians and other patient organisations from the ultra-rare community to discuss the extensive campaigning activity in recent months on access to treatments for patients living with ultra-rare diseases in the UK.

The panel consisted of Greg Mulholland MP, who has championed the campaign, and he was joined by Professor Chris Hendriksz, Action Duchenne's CEO Paul Lenihan and a parent of a young boy with Duchenne Muscular Dystrophy campaigning for the treatment Translarna, a drug caught up in the same access debacle as Vimizim for Morquio. Christine Lavery and Katy Brown, mum to six-year old Sam Brown who has led the campaign Keep Sam Smiling, spoke eloquently and powerfully and concluded the panel's speeches.

The event focused on the fact that Government has been explicit in its support for the life sciences industry and as a result the UK is largely considered a global location of choice for conducting research, development and clinical trials. But what good are these new treatments if they don't make it into the hands of those that need them most? A decision on the only life changing treatment available for this ultra-rare condition is always going to be emotional but months of uncertainty and constant process failures is unacceptable.

What makes this an even more intolerable situation and all the more painful is that 34 of these 77 children and young adults once had access to treatment via a clinical trial. They were pioneers of this new medicine. They and their families braved the stresses and the strains of five years of clinical trials, all in the name of medical innovation and lifesaving discovery.

What is critical is that this new medicine, Vimizim worked and was having a dramatic impact, greatly improving their health and their quality of life. By taking part in UK-based trials, these 34 children and young people made it possible for Vimizim to be licenced. In the last 14 months hundreds of other Morquio patients, in over 30 other countries, have and are accessing reimbursed Vimizim. This includes 176 people in Turkey and 3 in Greece. Why then, after their extraordinary commitment, is the NHS denying children and young people in England the same right?

Katy concluded her speech highlighting that not having a solution to deal with ultra rare diseases is deliberate and direct discrimination against a small group of vulnerable people who already have life and the system stacked against them. It is apartheid based upon the rarity of the condition that they were unlucky enough to be born with. If this were race, gender, age – it would wholly unacceptable and moreover illegal. Why is this any different?

The discussion that followed was hugely beneficial to all in the room. It was really encouraging to hear so many voices in support of getting fair access to treatments for rare diseases and we will keep fighting for the whole rare disease community.

Charlotte Roberts
Communication Officer

News From the Board of Trustees

The Society's Trustees meet regularly. Here is a summary of the main matters that were discussed and agreed at the Trustee Board Meeting on the 26 – 27 February 2015 at MPS House, Amersham

Treasurer's Report

Treasurer, Judith Evans, presented the financial report, cash flow sheet. It was agreed that the profit and loss figures are looking healthy. The Trustees scrutinised the accounts for year ending 31 December 2014. The Trustees expressed their gratitude to Gina Smith, Finance Officer and Mike Caputo, Auditor, for producing such a comprehensive set of accounts. The Trustees approved the accounts subject to the amendments agreed.

The Trustees approved the year end accounts of the dormant Company 'Wicked Genes' and these were duly agreed and signed.

Grant Awards

The following grant award was agreed:

£10,000 - Dr Brian Bigger, Genistein Clinical Trial

Access to New Therapies

Trustees were appraised of the campaign to secure reimbursed enzyme replacement therapy 'Vimizim'.

Policies

The Reserves Policy was approved unanimously

Trustees had been provided with a number of papers on the new SORP to read in advance of the meeting. The Trustees considered the advice relating to FRSSE SORP and FRS 102 SORP and agreed unanimously the Society should adopt the FRS 102 SORP in respect of the year end accounts for 31 December 2015.

MPS Commercial

Mahboob Khan presented his feedback from his first six months in post as Business Development Officer for MPS Commercial and his proposed strategy for the next six months including establishing brand identity, loyalty and legacy in order to consolidate the business and build on strong foundations.

WE NEED YOUR HELP

We are inviting all members to get involved with a new project that we are supporting, working with a company called FDNA (Facial Dysmorphology Novel Analysis).

As you are aware, many genetic diseases are associated with certain facial characteristics, and FDNAS have developed specialized software to analyse this in order to monitor disease progression.

If members would like to get involved to help test this software, they will be asked to supply up to 9 photographs of their children with MPS, aged 1 - 9 years.

You will find more details of this project, along with consent forms, enclosed with this magazine.

We would be grateful if as many of you as possible could support this worthwhile project.

What's On

MPS Regional Clinics 2015

MPSI - GOSH:
14th July
22nd September • 22nd December

MPSIII - GOSH:
8th September • 8th December

MPSIV - GOSH:
13th October

Fabry clinic - BCH:
23rd October

MPS clinic - BCH:
27th November

MPSI Post HSCT (over 6 years) - RMCH:
17th July • 16th October

MPSI Post HSCT (under 6 years) - RMCH
24th July • 23rd October

Conferences and Regional Events

Legoland Family Day
Sunday 9th August 2015

Childhood Wood Remembrance Day
23rd August 2015

Childhood Wood planting
25th October 2015

The Irish MPS Society are organising: All Ireland Conference - Clarion Hotel, Dublin
11th-12th September 2015

Northern Ireland Christmas Party - Dunsilly Hotel, Antrim
17th December 2015

Announcements

New Faces at the MPS Office

Emma Henry

Hi everyone, My name is Emma Henry and I have recently joined the MPS Society Team. I have been here for a few weeks now and am really enjoying my position as Fundraising Administrator.

This is the first time I have worked for a charity and in a fundraising role; however the team have welcomed me and made me feel at home already. I have learnt a lot about MPS Diseases in this short space of time, and I am looking forward to learning more and being able to help people that are suffering with these diseases, and their families.

My previous job roles have included administration and marketing roles for various companies which I hope will help me in this exciting new position. I decided to change my career path slightly and move into charity fundraising as I really wanted to feel I was helping people. My father has always worked with charities in disaster relief and I think this is where I have gained my passion for wanting to make a difference.

Most of my spare time is taken up by my animals, a horse and a dog that both keep me very busy. I enjoy keeping fit and healthy by walking, running, horse riding and attending the gym. I also love spending time with friends and family.

I am really looking forward to developing in my role and making a difference to the families at the Society.

Catherine Donald

Hello, my name is Catherine and I joined the MPS Society a few weeks ago as Finance Officer, to support the Clinical Trials Patient Access team.

I studied Accountancy at Aberdeen University and then went on to qualify as a management accountant. I've worked for several large organisations including Marconi, the Burton Group, Lidl and Equinix together with a few smaller companies. The MPS Society is my first charity and so I'm looking forward to broadening my experience. It's been a hectic few weeks familiarising myself with the various study trials and families on the trials – a far cry from counting tins of baked beans at Lidl!

Out of work, I'm a busy mum looking after two teenagers and a husband with a crazy work schedule. My youngest was diagnosed with Crohn's disease last year and the advice from the charity CICRA helped us enormously to get through 6 weeks of an elemental diet and I'm off to their IBD family information day in a few weeks.

The MPS Society similarly offers a huge variety of assistance and support and I can assure you that everyone here at MPS House is extremely dedicated and here to help you. I'm looking forward to being part of this great team.

New Members:

Darren & Amanda Scott have recently been in contact with the Society. Their daughter has a diagnosis of Sanfilippo Disease. Sophia is 4 years old. The family live in Scotland.

Glynis has recently been in contact with the Society. She has Fabry Disease. The family live in the Lincolnshire area.

Ms Webb has recently been in contact with the Society. Her daughter Samantha has a diagnosis of MPSIII. Samantha is 28 years old. The family live in the West of England.

Marc has recently been in contact with the Society. He has a diagnosis of Fabry Disease. The family live in the Cambridge area.

Judith has recently been in contact with the Society. She and her son Sean have a diagnosis of Fabry Disease. The family live in the Cleveland area.

Gintare & Donatas have recently been in contact with the Society. Their daughter Emma has a diagnosis of Alpha Mannosidosis Diseases. The family live in the London area.

Advocacy

The MPS Advocacy Support Service has been established since the Society was founded in 1982. At this time there were only 40 known families throughout the UK. The support provided was on a voluntary basis and depended heavily on individuals and parents to provide support to individuals diagnosed within their immediate and surrounding areas.

However in 1991, the Society opened its first office and with this the advocacy service we know today was born.

The MPS Society provides, through a team of skilled staff, an individual advocacy support service to its members. The service is flexible and a wide range of support is offered on a needs led basis.

The rarity of these conditions means that in many cases, accurate assessments, support and advice are not given due to the vast majority of social care and health professionals knowing very little if anything about the diseases.

Support provided by the team

• Telephone Helpline

0345 389 9901– the Society provides an active listening service, information and support. This includes an out of hours service

• Disability Benefits –

In understanding the complexities and difficulties individuals and families have in completing claim forms for Personal Independent Payment, the Society continues to provide help and support in completing these forms and, where needed, will take a representative role in appeals and tribunals

• Housing and equipment

– The Society continues

To take a major role in supporting and advocating appropriate housing and home adaptations to enable the needs of an individual with an MPS or related disease to be met. Where requested, we can provide comprehensive and detailed housing reports based on individual need

• Education –

The Society helps members to access appropriate education and adequate provision for its implementation. This is achieved through providing educational reports used to help inform and educate professionals, and in many instances, to inform Statements of Special Educational Need. Where requested, we also provide information days/ talks to schools and relevant professionals

• Respite Care –

The Society continues to work closely with a number of respite providers and, where appropriate can make individual referrals

• Independent Living/ transition –

The Society provides advice, information and support through the transition from child to adult services. This could include access to independent living, learning to drive, further education and employment

• MPS Careplans –

The Society undertakes a comprehensive assessment of the issues which need to be addressed when caring and providing support to a specific individual diagnosed with an MPS or related disease, as well as other family members through the writing of a careplan

• Befriender Service –

The Society links individuals and families affected by MPS and related diseases for mutual benefit and support

• Bereavement support.

For more information on any of the above or if there is anything else that you would like to chat with the advocacy team about please contact us:

Email: advocacy@mpssociety.org.uk
Telephone: 0345 389 9901

Advocacy Resources

The Advocacy Team have also developed a range of information resources focussing on particular issues which are available to download free of charge from the MPS website, www.mpssociety.org.uk

- Life Insurance
- Travel Insurance
- Hospital Travel Costs
- Disabled Access Holidays • Carers Legal Rights
- Carers Allowance
- Wheelchairs and Flights • Guide to Housing and Disabilities Facilities Grant
- Benefits including, Personal Independent Payment, Benefit Cap, Council Tax Benefit and Universal Credit

Each of our England based Advocacy Officers works with specific disease groups as listed. However, every member of the Advocacy Team has knowledge of all the diseases and may at times provide support in other areas dependant on need and individual assessment.

Team Members

SOPHIE

Manages the MPS Advocacy Team

STEVE

MPSIII Sanfilippo type A,B, C and D, MLD AGU, Winchester
Geleo Physic Dysplasia Sly, Gangliosidosis, Sialic Acid Disease

ALISON

Supports members living in Ireland

DEBBIE

MPSIVA Morquio, MPSI Hurler BMT, Hurler Scheie, Scheie, MPSVI Maroteaux-Lamy, MSD, MLII

REBECCA

Fabry
MPSII Hunter
MLIII / MLIV
Mannosidosis, Fucosidosis

Kidz South Event

On 4th June I attended the Disability Living Kidz event. This event has several locations over the UK now including Scotland and Wales. It is a free event for families and professionals. There is an exhibition of items such as bathing equipment, beds and wheelchairs and also other services such as education providers and charitable funding represented as well. I found it very informative and there were lots of families there trying out equipment.

There is also a programme of useful seminars which can give an insight into topics such as managing challenging behaviour and transition to adult services. These were well attended by a mix of professionals and family members and as well as gaining new knowledge were also an opportunity for questions too. I left the venue with lots of information and brochures.

Further details can be found on this website http://www.disabledliving.co.uk/Kidz/Welcome and there are the following events still to come: Kidz Scotland – 17th September 2015, Kidz to Adults North – 19th November 2015 and Kidz to Adults in the Middle – 10th March 2016.

Debbie Cavell
Advocacy Support Officer

Bereavements:

We wish to extend our deepest sympathies to the family and friends of:

John Paul O'Neil who suffered from MPSIII Sanfilippo disease and passed away on 13th April 2015 aged 36 years.

Natasha Pace who suffered from MPSIII Sanfilippo disease and passed away on 1st May 2015 aged 19 years.

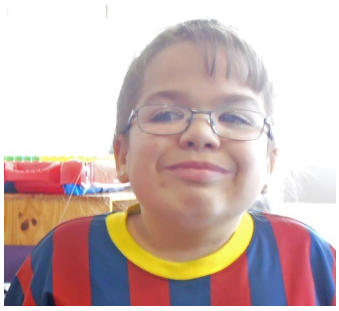
Jade McAfee who suffered from MPSIII Sanfilippo disease and passed away on 6th July 2015 aged 17 years.

Clinics

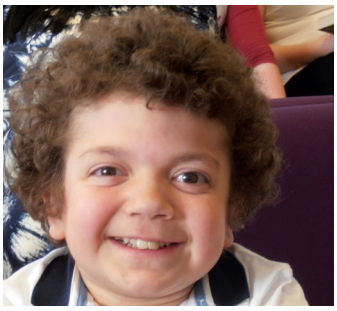
Recently the Advocacy Team have attended clinics at Great Ormond Street Hospital, Royal Manchester Children's Hospital and Birmingham Children's Hospital. It is always a pleasure to meet with the families and the clinical teams. It gives us a valuable opportunity to meet and chat with families face to face, but please remember that members can contact the Advocacy team at any time with queries or to share news.

Manchester MPSI BMT Clinic - 17th and 24th April 2015

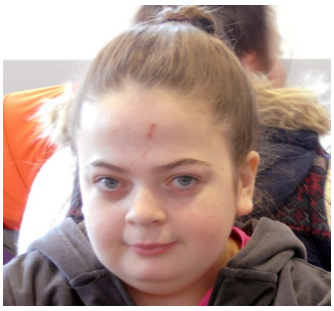
Alicia



Cody



Jamie



Rachel

Thomas

Ethan

Avah

Charlie

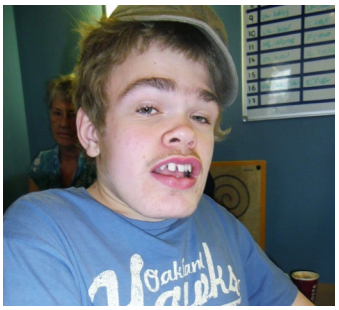
Birmingham Children's Hospital MPS Clinic - 8th May 2015

Roman

Sultan

Zena

GOSH MPSIII Clinic - 26th May 2015



Jamie

Birmingham Children's Hospital MPS Clinic - 12th June 2015

Emily & family

Isaac & family



All Ireland Advocacy Support Update

This last quarter has been a very different kind of busy for the All Ireland Advocacy and Support Service! Alongside all my usual Advocacy and Support work I have been busy working alongside professionals in Northern Ireland to organise a very special event.

On the 14th of May we held a very special meeting and celebration meal to mark the 20th Anniversary of the first MPS Specialist Clinic in Northern Ireland. This was an absolutely fantastic event and was well supported by both families and professionals from across Northern Ireland and beyond. We had a packed meeting programme with information about MPS identification and management, respiratory management, orthopaedic issues, the role of the MPS clinic and the MPS Specialist nurse and (of course) the role of the MPS Society.

The All Ireland Advocacy and Support Service is only 5 years old so I think that the words of Christine Lavery and Dr Fiona Stewart taken from our Anniversary meeting booklet say more than I ever could:

Alison Wilson
Advocacy Support Officer

Jibreel Arshad & Dr Fiona Stewart enjoying the celebratory dinner!

When we started the MPS Society in 1982 the goal was to provide a comprehensive support programme to all those affected by MPS and related diseases across the four countries of the United Kingdom. With close links to Northern Ireland, my late husband, Robin, was born and grew up in Belfast, we were determined that the MPS families in the Province would not be isolated by their condition or the political challenges that dogged the 1980s and 1990s. Throughout the 10 years up to 1993, I was a regular visitor to Northern Ireland as I worked on rare diseases for the disability charity 'Contact a Family'. During that time I met Dr Fiona Stewart and proposed the Northern Ireland MPS Clinic. The MPS Society was already holding expert Regional MPS clinics in other parts of the UK supported by Professor Ed Wraith who sadly died two years ago.

Twenty years ago Dr Stewart and I hatched a plan and the yearly (to become twice yearly) Northern Ireland MPS Clinic was born. Dr Ed Wraith supported Dr Fiona Stewart at these clinics for the next 15 years until sadly he became ill and Dr Simon Jones also from Manchester Children's Hospital succeeded him in the role. During the 'Troubles' the clinics were held at hotels before moving to Antrim Hospital some years later providing best care and clinical management of patients.

It has been a great pleasure to work with all the families in the Province over the years and special thanks to Dr Fiona Stewart MBE for her total dedication to the MPS community in Northern Ireland.

Christine Lavery
Group Chief Executive

Top photos, left to right: Dr Siobhan O'Sullivan (Metabolic Paediatrician), Dr Bronagh Sweeney (Metabolic Paediatrician), Jean Mercer (Metabolic Nurse Specialist) and Aidan Kearney (MPS Society member); Dr Alex Magee, Dr Fiona Stewart and Dr Tabib Dabir (all Consultants in Genetic medicine), Alison Wilson (MPS staff) and Siobhan Harding-Lester (Genetic Counsellor); Rab and Margaret Coyles and the Stewart family (MPS Society members)

Your Stories

Hannah's Children of Courage Award

Hannah Cooper, MPSI (post BMT), was very proud to receive a Children of Courage Award at the end of March. The Award ceremony was organised by The Rotary Club of Biggleswade Ivel and sponsored by local industry. Hannah was nominated for her award by Mrs Newman, Head of year 5 at Henlow Church of England Academy. Mrs Newman nominated Hannah for her positive attitude, perseverance, determination and good humour, despite the ongoing and tough challenges and difficulties that she faces as a result of her condition. Hannah was thrilled to receive her award from Cllr Caroline Maudlin - Chairman of Central Bedfordshire Council. Well done Hannah!

Nicola Cooper

Walking in Sunshine - an Expression of Hope artwork

Rachel recently contacted us to let us know that she and her daughter had submitted an artwork to Genzyme's Expression of Hope campaign, which gives those affected by rare diseases the chance to express themselves by producing some art. Rachel and Charlotte's picture, entitled 'Walking in Sunshine', has been shortlisted to be shown all over the world.

If you would like to know more about Genzyme's Expression of Hope campaign, please visit their website: http://expressionofhope.com/about/about-program

Charlotte (4) and I (39) painted this picture together and it truly encapsulates our journey with Fabrys disease. Since Charlotte and her dad's diagnosis 2 years ago, we have been on quite a journey with hills to climb and new scenery to see. We chose however, as a family, to embrace the life we have been given - to walk in the sunshine and try to forget about the rain. We chose to reflect on this as a blessing, to grab every moment by both hands and live life to the very fullest, whilst we can. We hope our painting reflects the path we have chosen to take on this unexpected journey

- Rachel

Being Positive - Archie's story so far

I remember standing in the kitchen with my wife Yvonne when we heard that Archie would be born 'special'. At that moment, life seemed to momentarily stop as we thought of the consequences for the future – what would the next few years hold for us as we rose to the challenge of caring for a child with MPSVI? For the next few weeks, Yvonne and I went through a true 'roller coaster' of emotions from sadness and self pity, through to disbelief – 'why us' was a phrase often stated.

However, whilst neither of us are particularly religious, we both found strength from believing that this was our destiny in life – to care for our little bundle of joy, come what may. With this strong belief, the negative feelings started to be overshadowed by feelings of strength, commitment and the desire to fight this condition.

"We feel touched to have been surrounded by so many angels"

We were fortunate. Archie was diagnosed within 9 months of birth, and within weeks we were off to Manchester Childrens hospital to start a new life with Ed Wraith and the Manchester Travel Lodge! To begin with it was quite fun – travelling up to Manchester on a Sunday evening, stopping off for tea on the way it was certainly a long week. I remember trying to heat up food on the way back down the M5, standing in the travel lodge car park in my pyjamas after a fire alarm and trying to eat dinner in our hotel room while being stared out by two blue eyes over the top of a travel cot - we have a lots of memories from the early days.

Being in hospital nearly every week has made us realise that many families are worse off than us - having a son that still manages to play a short game of football on a Saturday, go to Beavers and wrestle his daddy is just perfect. We have had to adapt to life with MPSVI – Archie no longer has a trampoline due to his weak neck (but now has a new fort he can climb on), when we wrestle I let him down gently on the floor so not as to hurt his back and when he plays football he has to have regular breaks. But life goes on and so have we.

Archie goes to a regular school and his friends are intrigued by his body brace (armour) and get quite frustrated with his teacher when she forgets that Archie has treatment day on a Wednesday. He is an important part of school life and his friends look after him – forming an important social network around him – supporting him through the dark times and making him laugh during the bright.

We cannot describe the support we have recieved from all of the surgeons, nurses and care assistants that have made a positive difference to our lives. We feel touched to have been surrounded by so many angels – least of all our family and friends that have supported us through the darkest days. However the biggest change to our life, which turned our dark future into one with a small flickering light was the first day we met a grey haired, warm hearted, pony tail wearing professor simply called 'Ed'. From that point onwards we had hope and continue to have it through the work his colleagues and associates continue in his honour.

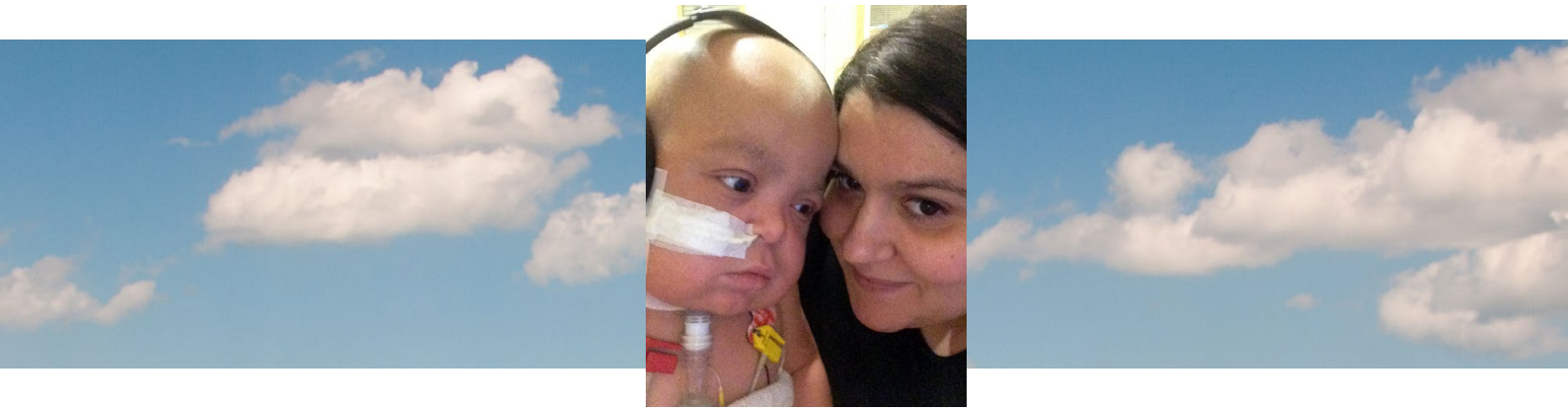
The MPS Society has inspired us to do more in our lives and in April this year, Yvonne and I, accompanied by some mad friends, will be running the 'Monster Race' to raise some money for the charity – let's hope we survive!

Archie is now 7 and doing well. His treatment has really helped him and we are full of hope for the future. We remain positive and, alongside the MPS Society, are determined to fight this disease until a cure is found.

Philip Pearson
Proud Father of Archie Jack Pearson

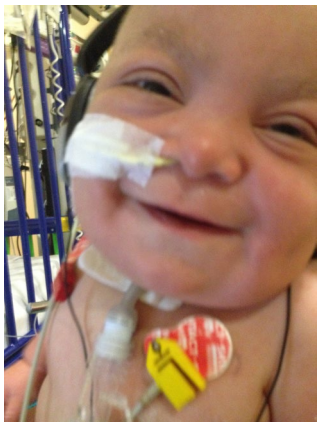
Thank you to Philip and his wonderful team who ran the Monster Race and raised a spectacular €2,037.50 on his Justgiving page.

Remembrance



This is our son, Max...

Max was born in November 2013 and seemed perfectly healthy to all of us. It was in the maternity ward that I knew something wasn't quite right with my new baby son. The very next day he failed his new-born hearing test and this worried me instantly. At times I could "hear" Max breathing. I raised my concerns with the medical staff who tried to reassure me that he was fine and had a little mucus from birth and it would settle. Once we were home, things didn't improve his noisy breathing worried both my husband and me and we took him to the doctor. I explained that it sounded as though Max had a really bad cold but he didn't have anything related to a cold. We also noticed during a routine nappy change that Max had a lump in his groin area. I immediately knew it was an inguinal hernia. So once again back to the doctor and she made 2 referrals; 1 to ENT for his noisy breathing and the other to paediatric surgery.



In April 2014 Max was admitted for his hernia repair. Alan and I kissed Max as he drifted off to sleep. We waited and waited. The 1 hour estimate we were given turned into 2.5 to 3 hours and I was getting anxious. Finally we were called back to the recovery room. He was fine. We were so relieved it was all over. Little did I know that it was only the beginning...

Once Max was back on the ward the Anaesthetist came to us and told us that everything went well apart from a little difficulty intubating Max for surgery and she asked the nursing staff to collect a blood and urine sample from Max for tests. It was simply explained to us that it was a metabolism test and nothing to be concerned about.

The next day a respiratory consultant came to meet me and asked me about Max's breathing issues. I asked him why and if he suspected cystic fibrosis. He said no, but arranged a genetics appointment. The geneticist looked at Max and asked me questions I had already answered before. She studied Max and looked at me and said from what I can see I know Max has MPS. MPS...??? I asked. She explained and that's when my whole world fell apart around me, my heart was thumping, his whole life flashed before my eyes, within minutes I was grieving for my baby boy, the loss of his health, his future and I blamed myself. What did I do? What did I not do? Did I eat something to cause this? My head was spinning, I felt sick and I was alone with Max and I looked at him and he smiled back at me and I asked. Why? why is this happening?? You see Max is oblivious to everything all he wants is cuddles, food and love. All he does is smile, eat and do what every other baby does. Looking at him he was perfect.

We started him on enzyme replacement therapy infusion once a week to try and give his body what he lacked. But we were told that though this infusion was very good at working locally in his organs it would not cross the blood brain barrier or affect his bones. So mental retardation was inevitable unless we allowed him to have a bone marrow transplant. We immediately agreed and started to make the necessary arrangements to start the process.

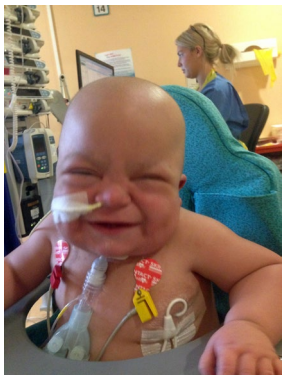
Alan and I tried to prepare for what lay ahead. Max and I would be away from home for up to a whole year. We have 3 other children which made it even harder. On the 18th August 2014 Max was admitted into the Oncology ward to start his chemotherapy in order to destroy his bone marrow to make way for his new cells.

After 8 days of intense chemotherapy sessions Max was ready for his new cells. On the 3rd Sept 2014 Max got his new donated cells. Then it was a waiting game to see if those new stem cells would graft and eventually replace his own bone marrow and produce the enzyme. It's a very long, slow process but I've been with Max every step of the way. He proved much stronger than I could ever be and I'm so proud of him. He was only 10 months old and he's had more surgeries, tests, drugs and hospital admission than I care to remember. But I prayed every day for him and hope that in a few weeks' time he will have 100% donor cells and be well enough to get out of isolation.

I only saw my husband and other children about twice a week. It's hard and not a day went by where the pain of

missing them reduced me to tears. Each visit was a disaster - because Max was in isolation he wasn't allowed anyone in the room apart from medical staff, Mummy and Daddy and his siblings would only see him from the other side of the room window. So Alan and I would swap children. He would stay with Max and I would go to see my other kids; meaning that Alan and I couldn't spend a minute with one another. It was a mere hug hello and a kiss goodbye. We did this for close to 6 months. It was awful.

Then sadly we were informed that Max transplant was unsuccessful. We were devastated. It forced us to make very hard choices. We had 2 options. We either give him back his own cells and let nature take its course with him or we repeat the procedure again.



We decided to give Max another chance. We asked for another transplant. Max was so sick; a cold landed him in PICU. I stayed with him the whole time. To top all that he still had a week of chemo to endure. It was the worse time of my life and I was helpless for him. All I could do was watch and pray for a miracle.

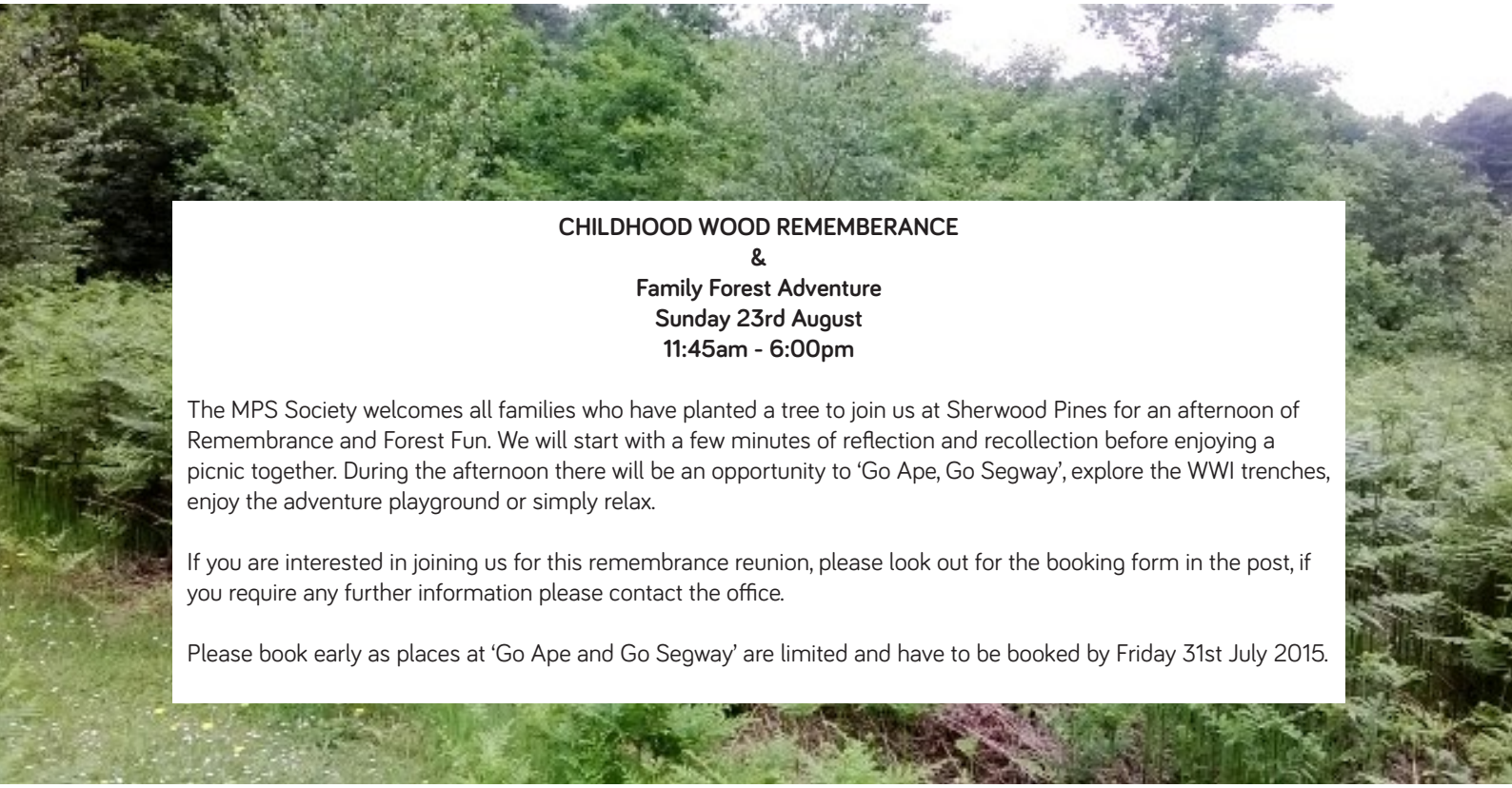
Miraculously Max made it through a second dose of intense chemo and then came the day for his second transplant. It was an exciting day. I was so optimistic. I had a great feeling about this time round. After 2 weeks we started to see the cells take. His counts were rising and Alan and I were ecstatically happy for him. He'd been through so much and this was his reward. Life seemed to be getting better and soon he was allowed visitors in the room. He was on the mend.

But one night I was woken up by a nurse. Max had deteriorated and was struggling to breathe due to a respiratory infection. Max was taken back to PICU and put on a ventilator to take away the effort of breathing for him. It was 10 days before Christmas 2014 and Max wasn't getting any better. When I saw Max laying there my heart exploded into a million pieces - my baby boy was fighting for his life and nobody knew why or what was wrong. Eventually the doctors found out that he'd contracted scarlet fever and it had attacked his lungs. Max's little body had had enough. He couldn't fight any more. He was sedated and unaware of what was happening and not in any pain.

My husband Alan stayed the whole time and I stayed in Clic Sargent House with my other children. Then one night I decided to go see Max. I kissed his cheek, stroked his face and told him how much I loved him and how proud I was of him. And I left. Alan was with him.

They were alone just him and his daddy and at 11.30pm on the 6th Jan 2015 - Max died peacefully in his dad's arms. Although our lives will never ever be the same again, Max lives on in our heart. I've never met such a strong and spirited character such as Max and he was beautiful inside and out. Incompatible with life but he was our son. We wish things were different we can't and won't ever forget Max. He's touched the life of many people globally through Facebook and other sites I've shared his story on and Max is our hero. Always and forever young. Our angel, Max Power...!!!

Angela Caukwell
Max's mummy



CHILDHOOD WOOD REMEMBERANCE
&
Family Forest Adventure
Sunday 23rd August
11:45am - 6:00pm

The MPS Society welcomes all families who have planted a tree to join us at Sherwood Pines for an afternoon of Remembrance and Forest Fun. We will start with a few minutes of reflection and recollection before enjoying a picnic together. During the afternoon there will be an opportunity to 'Go Ape, Go Segway', explore the WWI trenches, enjoy the adventure playground or simply relax.

If you are interested in joining us for this remembrance reunion, please look out for the booking form in the post, if you require any further information please contact the office.

Please book early as places at 'Go Ape and Go Segway' are limited and have to be booked by Friday 31st July 2015.

MPS Weekend Conference 2015

The MPS Society was delighted to have held our Weekend Conference, which took place Friday 26th June to Sunday 28th June. The Conference not only gave professionals and families the opportunity to listen to a variety of presentations (a few of which we have highlighted opposite), but also provided a chance for those affected by MPS to interact with each other and those working in the field.

Speakers ranged from medical professionals, who are involved in ground-breaking clinical trials, to parents who shared their experiences of the care and education of their child. We hope that everyone was able to come away with new information that will prove beneficial to both your professional or personal life. It was a priviledge to be able to host all the speakers involved.

Saturday night ended in a fabulous Gala Dinner, at which we were delighted to be able to present cheques to various research projects (see below), and then encouraged everyone to let their hair down and dance the night away! Our chocolate fountain and 'Selfie Booth' proved particularly popular - and we have the photos to prove it!

Our thanks to our speakers and everyone who attended that made the weekend so successful. We look forward to holding the next MPS Weekend Conference in 2017.

The MPS Society was delighted to be able to make grant awards to the following MPS and Fabry research projects.

Cheques were presented at the MPS Weekend Conference's Gala Dinner by Chief Executive, Christine Lavery and Trustee, Bryan Winchester.

£60,000 to Dr Brian Bigger, University of Manchester

A phase III, double blinded, placebo controlled clinical trial of high dose oral Genistein Aglycone in Sanfilippo Disease (NOTE This brings the total to £560,000 raised for this clinical trial. A further £40,000 still needs to be raised to reach our agreed grant award total of £600,000)

£10,000 to Dr Simon Jones, Manchester Children's Hospital

Immune Intolerance Induction with Methotrexate in Hurler Disease

£20,000 to Dr Derralynn Hughes, Royal Free Foundation NHS Trust

Genotype-Phenotype Relationships in Fabry Disease to stratify and understand heterogeneity using extended family pedigrees (cheque collected by Dr Uma Ramaswami on behalf of Dr Hughes).

The Shauna Gosling Trust donated matched funding of £34,000 to Professor Simon Heales, Great Ormond Street Hospital

MPS Antibody Pilot Project to be carried out between the Royal Manchester Children's Hospital and the Institute of Child Health, GOSH



Speaker Highlights

We have had some excellent feedback about the range of talks that were presented over the course of Saturday and Sunday, with topics including best practice paediatric and adult care, transitioning, palliative care, therapies and treatments, education and clinical trials.

Below are just a few of the highlights taken from a variety of presentations

Simon Heales

Role of Antibodies in Enzyme Replacement Therapy

- ERT may be produced and extracted from human, hamster or carrot cells
- ERT may be detected as foreign in patients with LSDs
- Antibodies to ERT can be generated and can inhibit the activity of ERT, preventing uptake into cells
- High levels of Abs (Antibodies) preceded changes in biomarkers
- Controlling Ab Response to ERT include: immunosuppressive regimen to cause tolerance, immunosuppressive drugs, positive effects on biomarkers
- Early knowledge of Cross Reactive Immunologic Material (CRIM) is beneficial

Julie Whann

Sensory Dimensions of Care

- Sensory integration impacts on our arousal levels, level of alertness, relaxation and emotional state and wellbeing.
- A child with sensory dysfunction may struggle to achieve the correct state of alertness
- Tactile dysfunction can involve avoidance (hypersensitivity) or sensory seeking (hyposensitivity) & can be treated with tactile toys & experiences
- Low proprioception is due to poor registration of sensory information & can be treated with resistance & weightbearing activities

Debbie Cavell

Changes to the Education System

- The Education, Health & Care Plan came into effect September 2014
- The EHC plan is for children aged up to 25 years old with additional educational needs based on learning or physical difficulties
- To apply for an EHC plan speak to the school's SENCO
- The local authority will assess whether additional support is needed that the school cannot provide
- If families do not agree with the decision they have the right to appeal
- Contact Debbie at the MPS Society for support



Thank you to our volunteers!

As always, a key feature of the MPS Weekend conference was the fantastic childcare programme assisted by our enthusiastic and experienced childcare volunteers.

We were lucky in having wonderful weather for most of the weekend and the outing to Drayton Manor Park on Saturday was a great success, with the volunteers helping all the children, both MPS affected children and their unaffected siblings, to get the very best from their visit. All the children had a wonderful time at the Park and returned to the hotel in the late afternoon with broad smiles on their faces and many exciting memories to take back home.

Some of the older MPS affected children and their siblings and young adults, accompanied by volunteers, were taken to a local bowling alley on Saturday evening; and the younger guests had their very own disco and crafty area.

The outing on the Sunday morning was split between the National Sea Life Centre and Cadbury World in Birmingham and was also a great success.

We asked the children if they had a good time, and here are some of their responses:

"It was awesome. I had so much fun"

"I liked sleeping in a posh hotel and I made some nice friends"

"It was amazing!"

"I had lots of fun with my volunteer - amazing!"

"It was perfect. I love it. Thank you"

"It was fun, can't wait for next time"

"This weekend was awesome!"

"I have enjoyed myself very much...thank you very much for taking care of us"

"I made two new friends"

"Amazing. Fantastic!"

"It was lots of fun. I would like to come again"

"I liked going on all the rides!"




Thank you Jenny for 32 years as a volunteer!

This year's Conference brought with it mixed emotions as it marked the final year of Jenny Bates' 32 years of volunteering. Jenny, who has seen the MPS Society grow from its inception, was presented with champagne and chocolates at the Volunteers' Dinner in appreciation for her many years of amazing support.

Soon after the Conference Jenny wrote in to our office:

"Thank you so much for the lovely gift of champagne and chocolates - and also my enormous double-bedded room, all to myself! - presented to me at last weekend's MPS conference. It was very kind of you, and I have many happy memories of my 32 years as a volunteer!"

Thank you, Jenny - your hard work as a volunteer has been invaluable and we wish you all the best for the future.

Jon Coombes - My Experience as a Volunteer

It was my first time volunteering at the MPS conference, but it most certainly will not be my last.

On Saturday morning we arrived at Drayton Manor Theme Park. It was great to see the young people enjoying everything the park had to offer, from Thomas Land, roller-coasters and a zoo featuring a Sumatran Tiger!

The energy and enthusiasm demonstrated by the young people was remarkable, the jokes and banter shared between the groups made for a very amusing day out.

On Saturday evening an older group of young people and volunteers put on our bowling shoes and needless to say competition was in the air. Back at the hotel the Lego Movie proved extremely popular with both the young people and the volunteers.

The impact left on me by volunteering at the MPS conference will last forever. The young people exhibited outstanding positive attitudes and engaged fully with all the weekend's activities, showing extreme levels of enthusiasm. I will be booking my place at the 2017 MPS conference ASAP!

Jon Coombes

Events

Scottish Family Day

The MPS society held a special event on 15th March for Scottish families at the Hilton Airport Edinburgh Hotel, all of which was made possible by the kind support of a grant from the Souter Charitable Trust.

The day was made more special by the fact that it was mothering Sunday, and the mums were given a balloon flower each to celebrate their special day.

The families all sat down to a tasty two course buffet and the families caught up with each other while the children ran around and played.

The entertainment was from the fabulous Gary Dunn who performed magic tricks and created some amazing balloon models from monkeys in a tree to fish on a rod - the rod didn't stand up to the waving around and had to be repaired several times! The adults and children all loved the magic show and before we knew it, it was time to say goodbye.

Thanks to the Souter Charitable Trust and all those who attended. These events allow our Scottish families affected by MPS and related diseases to meet one another and to know that they are not alone, making new friends and meeting old ones and having lots of fun and laughter along the way.

Rebecca Brandon
Advocacy Support Officer

Scottish Family Weekend

On 10th - 12th April, the MPS Society held a Scottish Family Weekend at Sundrum Castle near the beautiful Ayrshire coast, thanks to a generous grant from The Adamson Trust.

Each of our families enjoyed their own caravan within the scenic grounds, which had been adapted where necessary for disabled access. Over the course of the weekend, the families made the most of the wide variety of activities on offer, including an indoor pool, an adventure playground, soft play area, mini tenpin bowling and evening entertainment. The nearby beach also offered the chance for the families to relax and unwind.

Our thanks to The Adamson Trust for funding this wonderful weekend break.

International

EuropaBio's Event on the Benefits of Biotechnology 23 June 2015, European Parliament Brussels

This meeting chaired by Carlo Incerti from Genzyme Sanofi was to showcase examples of benefits that biotechnology brings to people's everyday lives and covered agriculture and feed, industrial, environmental and healthcare. The healthcare pitch was given by Elcin Ergun, Executive Vice President, Merck Serono. My role was to give the 'Patient's Perspective' and it was a pleasure to use tangible examples of our member's experiences and benefits of being on enzyme replacement therapy. This was the first time a lay person had been invited to speak at this meeting and it was evident that up until this moment many people working in the healthcare sector of the biotech industry had had little exposure to the impact of their efforts on the patient.

The meeting also afforded me a platform to address the EU Commissioner for Health and Food Safety, Mr Vytenis Povilas Andriukaitis in respect of the failure of our Health and Innovation Minister, George Freeman, to find transparent and equitable processes for people with ultra orphan diseases like Morquio, MPSIVA, to receive reimbursed new therapies.

Christine Lavery
Chief Executive

NZORD
because rare disorders need understanding and treatment

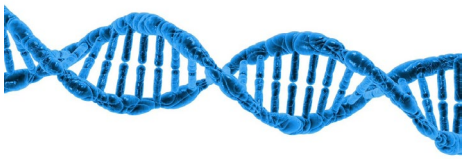
Progress in funding of orphan drugs in New Zealand?

It's a risky thing to predict the future, but based on recent informal discussions with Pharmac and with some pharmaceutical suppliers, it seems likely that in the next month or so, there will be arrangements made for a small number of orphan drugs to be added to the NZ pharmaceutical schedule and publicly funded. It has been a very long campaign to get this need addressed. Thanks to those who have worked with me, and not given up over many long and difficult years, to get this progress.

It will be a significant step forward to get new orphan drugs listed on the schedule for the first time in this century, in New Zealand. Pharmac and the politicians involved should be acknowledged for finally listening and responding to our case, especially in light of unreasonably restricted budgets they have been forced to manage with. The challenge that will flow from this will be the need to then allocate sufficient funds to do the whole job properly for the rest of the rare disease population who are waiting for their treatment to be funded.

John Forman
Executive Director
New Zealand Organisation for Rare Diseases

Research & Treatment



Gene therapy for MPSII

Mucopolysaccharidosis type II, also known as Hunter disease, is a lysosomal storage disorder caused by mutations in the iduronate-2-sulphatase (IDS) gene. These mutations alter the production of the IDS protein that degrades complex sugars, or glycosaminoglycans (GAGs), found throughout the body. Due to this malfunction, long chains of sugar molecules known as heparan sulphate and dermatan sulphate accumulate in the cell's waste disposal system; the lysosome. This progressive accumulation of GAGs in cells means that over time, cells become damaged and symptoms start to appear. Currently, MPSII is subdivided into two phenotypes; attenuated or severe, although the spectrum of severity is remarkably wide.

Patients with a more severe phenotype experience developmental delay, cognitive decline, airway and cardiac disease, as well as skeletal problems, whilst attenuated patients only retain the systemic symptoms without CNS involvement. Current treatment options for MPSII are focused on improving the patient's quality of life. The only approved treatment is enzyme replacement therapy (Elaprase®, Shire) where the missing enzyme is infused back into the patient through intravenous infusion. Although encouraging for treating somatic symptoms, Elaprase® is unable to cross the blood-brain barrier as it prevents large molecules and toxins from entering the brain. Hence treating the CNS remains the most challenging part of therapeutic development.

Ideally, the replacement of the malfunctioning gene with a functioning copy in cells would provide a very effective therapy. As such, gene therapy approaches take advantage of viruses that infect human cells and are engineered to contain the missing gene, whilst removing the viruses' own DNA, to create gene therapy vectors. Adeno-associated viruses (AAVs) are small viruses that can infect both dividing and non-dividing human cells without causing disease.

When AAV is engineered to contain the missing gene and administered intravenously, a very high number of viral particles are required to effectively treat the somatic organs. This approach is particularly costly and high doses of AAV are likely to generate antibodies against the AAV itself, reducing its therapeutic effect. Most AAVs are unable to cross the blood-brain barrier from the blood, therefore unable to adequately treat the CNS.

To bypass this issue, intracranial AAV delivery is used to inject AAV into the brain and deliver viral particles to resident brain cells. Whilst a very powerful tool for treating the CNS, intracranial AAV delivery is held back by scale up issues and uneven or insufficient distribution of virus particles throughout brain tissue. Indeed, most pre-clinical studies are performed on mice brains that are small in volume (0.5cm3) compared to human brains (1200cm3).

Our current therapeutic approach, known as stem cell gene therapy, uses a lentivirus vector containing the missing IDS gene. This virus can infect bone marrow that is harvested from the patients themselves, treated outside the body with the virus and infused back into the patient. This corrects haematopoietic stem cells that make up the blood and provide the body with a correct version of the IDS gene. Cells in the bone marrow are able to differentiate and migrate to the brain where they cross the blood-brain barrier to act as reservoir cells that secrete the enzyme for other brain cells to take up. Ex vivo gene therapy heavily relies on the transduced cells themselves giving the expansion that is lacking with AAV therapy. We are currently developing an ex vivo gene therapy treatment for Hunter in our MPSII mouse model, taking advantage of our expertise with this approach in MPSIIIA.

Hélène Gleitz

Stem cell Gene Therapy for MPSIIIA and B

MPSIIIA patients have a deficiency in the Sulfamidase enzyme. This leads to the build up of partially degraded sugars called glycosaminoglycans (GAGs) to toxic levels in the cells of the body with the brain most affected. The Stem Cell & Neurotherapies group at the University of Manchester are developing a new therapy for MPSIIIA that boosts the level of missing enzyme in patients' own cells. The gene for the missing enzyme is delivered using a lentiviral vector to patient stem cells isolated from their blood and these corrected cells are then transplanted back into the patient. The cells repopulate the immune system and start to produce active enzyme in the blood that can be taken up by all cells in the body, reducing the amount of toxic glycosaminoglycans (GAGs) stored.

The enzyme produced cannot pass into the brain but some cells from the bone marrow are able to move into the brain to release enzyme which can then be taken up by brain cells. In a MPSIIIA model of the disease, GAG storage and inflammation in the brain are corrected to near normal levels following treatment with 11% normal enzyme levels being produced. The treatment is now in the latter stages of development with final pre-clinical safety and efficacy testing underway. A clinical trial for this new therapy is planned to begin in early 2016. In addition, a similar stem cell gene therapy approach is currently in development for MPSIIIB at the University of Manchester with promising results so far in a model of MPSIIIB disease.

Brian Bigger

Amicus: More Good News on Migalastat

Study patients show continued kidney stability

Amicus Therapeutics announced in October additional positive data from a Phase 3 study (Study 2011) of the oral small molecule chaperone migalastat HCl ("migalastat") in Fabry disease patients with amenable mutations.

Dr. Daniel Bichet, Full Professor and Section Head, Renal Function & Transport Physiology, University of Montreal, said, "Baseline proteinuria levels are among the most predictive indicators of disease prognosis and kidney function decline in Fabry patients. The data presented today show that when comparing patients with similar levels of proteinuria, patients treated with migalastat are more stable in their kidney function versus untreated patients. These results are very encouraging for migalastat as a treatment for Fabry patients with amenable mutations."

Stratifying patients for gender and baseline proteinuria demonstrated that patients treated with migalastat experienced less decline in kidney function than untreated patients from a previously published natural history study.

"The additional data on substrate reduction show that we can accurately identify patients who may benefit from migalastat. We look forward to meeting with regular agencies starting this quarter as we work to make migalastat available for all amenable Fabry patients as quickly as possible," stated Dr. Jay A. Barth, Chief Medical Officer of Amicus.

With new data, Amicus prepares to market migalastat

Amicus is now preparing to submit marketing applications for migalstat as a monotherapy for Fabry patients who have amenable mutations.

Positive Phase 3 data in both treatment naïve and ERT switch patients have shown that treatment with migalastat has resulted in reductions in disease substrate, stability of kidney function and improvement in a key parameter (left ventricular mass index, or LVMI) in patients with amenable mutations.

For all other Fabry patients who do not have amenable mutations and cannot take monotherapy, Amicus is advancing migalastat in combination with ERT.

Anticipated 2015 Fabry Franchise Milestones:

- FDA meeting to discuss migalastat monotherapy in 1Q15
- Migalastat monotherapy MAA submission in mid – 2015
- Initiation of longer-term Phase 2 study of oral migalastat co-administered with currently marketed ERTs in 2H15
- Internal development underway of next-generation ERT) (bio-better Fabry ERT cell line for co-formulation with migalastat)

Research in Emerging Treatments

MPSI

Amicus Therapeutics disclosed a preclinical Chaperone- Advanced Replacement Therapy (CHART TM) program for MPSI on June 25, 2013. Amicus is developing a proprietary human recombinant IDUA (rnIDUA) enzyme co-formulated with a novel pharmacological chaperone as a next-generation therapy for MPSI The pharmacological chaperone is designed to improve tissue uptake and reduce the immunogenicity of rhIDUA by stabilizing the enzyme in its properly folded and active form. This next-generation enzyme replacement therapy is in the preclinical stage of the development pipeline. For more information visit http://www.amicusrx.com/.

Immusoft's mission is to develop a completely new platform for delivering medicines-programming a patient's own cells to become miniature drug factories. By instructing a patients cells to constantly secrete gene-encoded medicines (biologics), their technology would enable treatments that are currently infeasible due to short half-life , instability , or production and distribution challenges – all of which are significant limitations of current delivery modalities.

Immune system Programming (ISP TM) technology is based on an in vitro human B lymphopoiesis culture system developed in the Lab of Dr. David Baltimore at the California Institute of Technology (Caltech) and a novel lentiviral pseudotype discovered in Germany and developed in France. Immusoft has filed for an international patent on this novel combination of technologies and has exclusive rights to the components from Caltech.

The goal of ISP TM technology is to produce long-lived, biologic-producing plasma cells froma patients resting B cells. The approach would involve harvesting of B cells from a simple blood draw, which would then be genetically modified and differentiated into plasma celss in vitro. These plasma cells, which naturally have robust protein production capabilities, would then be injected back into the patient where they will home to the bone marrow, take up survival niches , and produce the intended therapeutic. A suicide gene system is added to the programmed cells, allowing for their rapid elimination if the therapy is no longer needed.

A clinical trial in patients with MPSI is being planned for early 2016 at the University of Minnesota. For more information visit www.immusoft.com.

Pentosan polysulfate (PPS) update on the development of therapy for MPS provided by Dr. Calogera Simonaro and Dr. Edward Schuchman.

Two studies have now been published using the rat model of MPSVI (Plos One , 2012 , 2014). These studies compared the effects of daily oral administration of PPS to once weekly subcutaneous (under the skin injection). Significant improvements were observed in both studies, including a reduction in inflammation, improved mobility and improved dentition and tracheal deformities. Notably, they also found that weekly subcutaneous administration significantly reduced GAG levels in tissues and urine of the treated rats.

Based on these findings in the MPSVI rats, a similar comparative study was undertaken in MPSI dogs. This work , which was ongoing for over 1 year, is currently being prepare for publication, but confirmed the safety and GAG-reducing effects of subcutaneous PPS treatment, and also revealed positive effects on MPS cardiovascular disease.

PPS is approved and marketed as an injected drug in serval European countries for various disease indications. To advance the development of PPS for MPS patients, two small investigator-initiated studies have been undertaken in Germany and Japan in adult MPS individuals (types I and II , respectively. The goal for these studies is to establish the safety of injectable PPS in adult MPS patients. Several clinical and biochemical endpoints also will be observed. Based on the outcome from these adult studies, safety studies in MPS children will be undertaken. We are anticipating that results from these two studies will be available in the first half 2015.

We are also pleased to report that several contractual agrements have been put in place to advance the clinical development of PPS for MPS. First, Mount Sinai will license relevant patents to **Plexcera Therapeutics**, a small drug company formed in 2013 based on research from their laboratory and for the advancement of therapies for rare diseases. Plexcera also has established an exclusive supply agreement with bene pharmaChem (Germany), the company who developed PPS and the only drug company that currently manufactures PPS approved for human use. In addition, orphan drug status for MPSI has been obtained by Plexcera from EMA and FDA. Orphan drug status for MPSVI also has been obtained from FDA.

Based on the outcome of the ongoing studies in Germany and Japan , during the upcoming year Plexcera will engage international regulatory authorities for advice on the most appropriate clinical development path going forward.

PPS is an example of one such therapy. For more information visit http://www.plexcera.com/.

PTC Therapeutics, INC. announced December 19, 2014 that both the U.S. Food and Drug Administration (FDA) and the European Medicines Agency (EMA) have granted orphan-drug designation (ODD) to Translarna (ataluren) for the treatment of patients with Mucopolysaccharidosis I (MPSI). There is significant unmet medical need and new treatments targeting the underlying cause of the disease are needed. MPSI represents the third indication for which Translarna has received orphan-drug designation. Translarna also has orphan-drug designation for Duchenne muscular dystrophy and cystic fibrosis.

Translarna is a protein restoration therapy designed to enable the formation of a functioning protein in patients with genetic disorders caused by a nonsense mutation. In preclinical models of MPSI , Translarna demonstrated that it crosses the blood-brain barrier and penetrates skeletal and cardiac tissues. The same preclinical models indicate that Translarna treatment reduces GAG levels in multiple tissues with no observed signs of cells toxicity or stress. Since Translarna is administered systemically, crosses the blood-brain barrier and penetrates other tissues relevant to disease, there is potential for Translarna treatment to address the cardiac and neurological defects associated with nonsense mutation MPSI.

Translarna, discovered and developed by PTC Therapeutics, Inc., is a protein restoration therapy designed to enable

the formation of a functioning protein in patients with genetic disorders caused by a nonsense mutation. A nonsense mutation is an alteration in the genetic code that prematurely halts the synthesis of an essential protein. The resulting disorder is determined by which protein cannot be expressed in its entirety and is no longer functional, such as dystrophin in Duchenne muscular dystrophy. Translarna is licensed in the European Economic Area for the treatment of nonsense mutation Duchenne muscular dystrophy in ambulatory patients aged five years and older. For more information visit htpp://www.ptcbio.com/.

ReGenX Biosciences is developing GXC-111 for MPSI Hurler disease, which involves a one-time delivery of a normal copy of the IDUA gene to cells of the central nervous system , allowing the body to produce enough enzyme to ameliorate symptoms without the need for repetitive treatment. A study performed by researchers at the Univeristy of Pennsylvania demonstrated that one-time delivery of RGX-111 can be accomplished *in vivo* and globally correct central nervous system manifestations of Hurler syndrome. The results, which were published in the journal *Molecular Therapy* support their planes to developRGX-111.

ReGenX is developing a new class of personalized therapies based on its proprietary NAV® Technology platform for a range of severe diseases with serious unmet needs, including lysosomal storage disorders. NAV Technology includes novel AAV vectors AAV7, AAV8, AAV9 and AAVrh10. For more information visit http://www.regenxboi.com/.

MPSII

ReGenX Biosciences developing RGX-121 for MPSII Hunter disease, which involves a one-time delivery of a normal copy of the gene encoding I2S to cells of the central nervous system, allowing the body to produce enough enzyme to ameliorate symptoms without the need for repetitive treatment.

ReGenX is developing a new class of personalized therapies based on its proprietary NAV® Technology platform for a range of severe diseases with serious unmet needs, including lysosomal storage disorders. NAV Technology includes novel AAV vectors AAV7, AAV8, AAV9 and AAVrh10. For more information visit http://www.regenxboi.com/.

MPSIIIA/B

Abeona Therapeutics, Inc. is developing gene therapy based potential cures for Sanfilippo (SF) syndrome Type A and Type B. In SF disease, the predominant symptoms occur due to improper cell function within the central nervous system(CNS), which result in cognitive decline, motor dysfunction, and eventual death.

They have two lead products , ABX-A an ABX-B, to deliver the therapeutic product to the CNS with the aim of reversing the effects of the genetic errors that cause the disease. ABX-A and ABX-B induce cells in the CNS and digestive tract. Safety studies conducted in large animal models have demonstrated that delivery of ABX-A and ABX-B are well-tolerated with minimal side effects. Importantly, efficacy studies in animals with Sanfilippo disease have demonstrated unprecedented therapeutic benefit months after treatment. A single dose of ABX-A or ABX-B significantly restored normal cell and organ function and increased the lifespan of animals with Sanfilippo disease over 100% a year after treatment, compared to untreated control animal. Similarly, animals treated with ABX-A and ABX-B demonstrated significant corrections of cognitive defects that remained months after drug administration.

These results are consistent with studies from several laboratories suggesting AXB-A and ABX-B treatment could potentially benefit patients with Sanfilippo disease Type A and B, respectively.

For more information visit http://www.nationwidechildrens.org/center-for-gene-therapy.

MPSIIIA

ReGenX Biosciences is teaming up with **Laboratorios Dr. Esteve** to develop and commercialize treatments for the rare lysosomal storage disease mucopolysaccharidosis type IIA (MPSIIAS, or Sanfilippo syndrome Type A) using ReGenX' NAV r AAV9.vectors for treatment of MPSIIA in humans.

ReGenX has given Esteve a nonexclusive worldwide license to the vectors per the agreement, along with rights to sublicense. In return, ReGenx will receive an up-front payment, milestone fees, and royalties on net sales of any product incorporating NAV rAVV9.

NAV is, according to ReGenX, a gene delivery technology that includes recombinant adeno-associated viral (rAAV) vectors such as rAVV7 , r AAV8 , rAAV9 and rAAVrh10. NAV rAAVrh10 was the focus of a similar agreement back in December between ReGenX and gene therapy firm Lysogene, also to develop and commercialize treatments for MPSIIIA.

MPSIIB

BioMarin Pharmaceutical Inc. announced December 10, 2014 that the Food and Drug Administration (FDA) has granted orphan drug designation for BMN 250, a novel fusion of alpha-N-acetygluosaminidase (NAGLU) with a peptide derived from insulin-like growth factor 2 (IGF2), for the treatment of Sanfilippo Syndrome Type B or Mucopolysaccharidosis IIIB (MPS IIB). BioMarin expects to initiate clinical studies with BMN 250 in mid-2015.

Discovered by BioMarin, BMN 250 is an enzyme replacement therapy using recombinant human NAGLU with an IGF2, or Glycosylation Independent Lysosomal Targeting (GILT) tag. BMRN 250 is delivered directly to the brain using BioMarin's patented technology. For more information visit http://bmrn.com.

MPSVI

The MeuSIX consortium plans to conduct a multicentre phase 1/2 clinical trial to investigate the safety and efficacy of AAV-mediated gene therapy in patients with MPSVI (Maroteaux-Lamy syndrome). An orphan drug designation (ODD) has been obtained from both the European Medicinal Agency and the US Food and Drug Administration for the MPSVI therapeutic AAV vector.

Pre-clinical studies have demonstrated that a single intravascular administration of an adeno-associated virus (AAV) encoding ARSB results in levels of expression of therapeutic ARSB for at least four years post injection and significant improvement in biochemical, visceral and skeletal features.

The results from this clinical trial proposed by the MeuSIX consortium has the potential to have a tremendous impact on the natural history of MPSVI and to significantly improve the quality of life of the affected patients. Moreover, the approach developed may facilitate the development of similar approaches for other inborn errors of metabolism. For more information visit http://meausix.tigem.it/.

Healthcare at Home's first patient involvement meeting

In close alignment to Healthcare at Home's (HaH) focus of building a truly patient-focused organisation, the first Patient Involvement Meeting took place in April in London. Members of the HaH executive team met with a group people currently receiving HaH homecare services to better understand how they could communicate more effectively with the people they serve.

The meeting, which was chaired by Elaine Strachan-Hall, Clinical Director at HaH, began with three presentations from the HaH executive team, focusing on current operations and the vision for the future. A workshop followed, where the attendees brainstormed ideas on what homecare means to them and how they believe HaH should better communicate with patients moving forward.

The key feedback from the patients who attended included:

• It would be useful for HaH to develop a more personalised complaints response process to ensure people feel assured that a 'real person' is dealing with their issue if something goes wrong

• A quarterly patient newsletter would be a useful way to receive company updates, especially if distributed with deliveries and during nurse visits

• A suite of information would be useful upon registering for homecare services with HaH, including; posters, leaflets and a booklet that provides FAQs and all contact details

This is just some of the feedback the HaH executive team are now scoping to implement for patients.

If you receive homecare services from HaH, and wish to have a say in the way this is delivered, please let us know. There are further meetings planned this year and your input would be valued.

Please contact the MPS Society on 0345 389 9901 to register your interest for involvement in future HaH patient activities.

Healthcare at Home have made some key operational improvements over the past month to improve their services, and they have updated us with the following information:

Improved complaints-handling process

A new system has been introduced that allows for simple complaints to be dealt with and resolved at the first point of contact, in customer services, without the need for escalation. This has enabled the team to more effectively manage complaints that require investigation and resolution.

Complaints related to patient safety are immediately escalated to the Chief Pharmacist and Patient Safety Team for investigation, resolution and prevention. Over the last month, 39.7% of simple complaints were resolved immediately at the first point of contact.

Reduction in number of urgent deliveries

Our medical delivery services are designed to ensure that patients have a 'buffer' supply of medicines to mitigate the risk of running out. However, if a patient does happen to run out of their supply, the same-day/urgent delivery service enables us to respond quickly.

As a consequence of improving our processes, from administration through to prescribing and dispensing, just 5-6 urgent deliveries now have to be made per day (0.14% of all daily orders) to meet patient needs.

Queen Mary University of London

Barts and The London School of Medicine and Dentistry

Medical Students: A tough audience for rare diseases

Joint co-authors: Debra Fine[1], Hannah Grant[1], Emma Keohane[1], Lucy McKay[1], Pat Roberts[2], Victoria Ward[1]

[1] Barts and The London School of Medicine and Dentistry, London

[2] Save Babies Through Screening Foundation UK

Background

- 1 in 17 people in the UK will be affected by a rare disease in their lifetime
- One of the main challenges is reaching a diagnosis
- Over 50% of medical students become GPs, the first contact for most patients, so it is critical that they recognise the signs of rare diseases
- The current undergraduate curriculum has limited teaching on when to suspect a rare disease

Methodology

- Our aim is to encourage medical students to develop **understanding and awareness** of rare conditions to reduce time to diagnoses
- Establish Barts and The London Society for Rare Diseases
- Hold specialist evening lectures and national symposiums
- Provide a variety of speakers including clinicians, patients and advocates
- Design a website to provide key information about the society and events
- Expand nationally by creating societies at medical schools across the UK

Conclusion

- The project has raised awareness of rare diseases to **future generations of clinicians**
- Positive feedback from medical students attending events has validated the success of our events
- To date rare disease societies have been set up in all five London medical schools
- Students are accessing resources via the website
- S4RD aims to encourage all UK Medical Schools to **adopt this model** by 2016

'REMEMBER TO THINK OUTSIDE THE BOX'

Results

October 2011
Founded Barts and The London Society for Rare Diseases

Students4RareDiseases (S4RD) is a national organisation supported by the UK LSD Collaborative patient advocacy group.

UK LSD Collaborative winning the prestigious Genzyme Patient Advocacy Leadership Award has enabled us to extend the concept to other medical schools.

PAL Awards Patient Advocacy Leadership Awards

April 2014
Second National Symposium
Held at Royal Society of Medicine in collaboration with the Medical Genetics Section:

- Attended by over 50 delegates
- Topics included MPS, primary lymphoedema and SCID

2011 — 2012 — 2013 — 2014 — 2015

January 2012
Launch of regular evening talks

- Ehlers-Danlos syndrome
- Paraneoplastic syndromes
- Fabry Disease
- Newborn Screening and Krabbe disease
- Retinoblastoma
- Batten Disease

February 2013
First National Symposium
Held at Barts and The London Schoolof Medicine and Dentistry

- Attended by over 40 delegates
- Topics included paediatric oncology, inherited skin disorders and relapsing polychondritis

2013
Founded S4RD
'Dare to think rare'

February 2014
Featured in The Independent national newspaper


8 · FEBRUARY 2014 A SUPPLEMENT BY MEDIAPLANET DISTRIBUTED WITHIN THE INDEPENDENT

NEWS

Training a future generation of doctors to diagnose rare diseases

A unique collaboration between a group of medical students and a patient advocacy group working to raise awareness of rare diseases amongst trainee doctors, has won a prestigious pharmaceutical industry award.

25th February 2015
Third National Symposium to be held at at RSM

LSD PATIENT COLLABORATIVE

www.students4rarediseases.org

Information & Resources

Patient Advocates for Newborn Screening is looking for a midwife to strengthen its Board

Save Babies UK provides support for those affected by Krabbe disease and works to advance Newborn Screening for metabolic diseases in the UK through its working group the Patient Advocates for Newborn Screening (PANS)

The working group, which is chaired by Save Babies UK, includes Climb (Children Living with Inherited Metabolic Diseases), The MPS Society, (Society for Mucopolysaccharide Diseases), AGSD UK (Pompe Disease) and ALD Life. The group is supported by health and diagnostic professionals including a Pompe Family Support Practitioner.

The group considers all issues surrounding New Born Screening in the UK, both generic and those in relation to specific diseases. The group work to identify and agree on diseases that should meet the criteria for submission to the UK National Screening Committee (UK NSC) new born screening programme and make recommendations and arguments in terms of those diseases following specific criteria within the terms of reference.

Patient Advocates for New Born Screening (PANS) is interested in appointing a Midwife representative to strengthen its Board which meets 4 – 5 times a year in Manchester Children's Hospital. This is a voluntary position with reasonable travel expenses reimbursed.

If you are a midwife with an approved midwifery degree to practice in the United Kingdom and are interested in this position please send an expression of interest on no more than 2 sides of A4 to:

Pat Roberts
Save Babies Through Screening Foundation UK
22 Bransdale Avenue
Leeds LS20 8QA
patroberts@savebabiesuk.org

Previous page shows a poster designed by Students 4 Rare Diseases

Students 4 Rare Diseases was founded by a group of 3rd year medical students at Barts and the London Hospital to create a learning module for students on rare diseases. The initiative came from the Lysosomal Storage Disease Patient Organisation Collaborative of who the MPS Society is a leading member. The LSD Collaborative supported the Students 4 Rare Diseases to successfully apply for a PALS Award from Genzyme in late 2013.

The Second Unusual Suspects Symposium organised by representatives from four London Medical Schools was held at the Royal Society of Medicine in April 2014 with 51 medical students attending. Organised by representatives from 4 medical schools. The key aim was to bring future doctors and rare disease patients together to emphasise the importance of diagnosis and appropriate management.

In September 2014 Students 4 Rare Diseases won the best poster prize (shown on previous page) at the International Symposium on Newborn Screening at the Regional European Meeting in Birmingham.

Boudoir Wine Bar in Glasgow was just one of many places to go blue for MPS Awareness Day.

Read more on page 35.

Fundraising

First of all a huge thank you to everyone who got behind our Wear It Blue and #Confess4MPS campaigns on or around MPS Awareness Day - as usual we had a terrific response and we are absolutely delighted that this special day is getting bigger and bigger each year! We have picked just a selection of the many fundraising events you held to mark the day, and these start on page 34.

So what now? Our supporters do not seem short of fundraising ideas of their own, but if you are looking for an idea why not try organising your own Wicked Walkabout? A sponsored walk is a great way to get some fresh air, spend some time with friends and family, as well as raise some awareness and funds. If you would like more information turn to page 46. We have lots of free fundraising materials that you can make use of, as well as some fantastic merchandise which is available from our online shop - just visit www.mpssociety.org.uk.

If you do hold a fundraising event, please don't forget to let us know what you have planned, either by dropping us an email (fundraising@mpssociety.org.uk), giving us a call (0345 389 9901) or even messaging us on Facebook or Twitter. We do like to send everyone who have fundraised for us a thank you letter, so please remember to let us know if you have paid in your donation as a bank transfer - or if you post us a cheque, a good old-fashioned note will be much appreciated!

Elkie Riches
Fundraising & Information Officer

Chiara's Clan do the Kiltwalk

The Kiltwalk is a fantastic Scottish charity which brings together some of the nation's best-loved childrens' charities in great fundraising events, taking place in both Scotland and overseas.

Michelle Petersen, mum to Chiara Fox (MPSI Hurler) and a group of family and friends took on Glasgow's Kiltwalk in support of the MPS Society. Read on for her story.

At 8 months old Chiara Fox was diagnosed with MPSI Hurler. Since that day Chiara has been an inspiration to everyone that has come in contact with her. She has touched the hearts of so many people and refuses to let anything get her down. When Chiara was diagnosed, as a mum I wanted to know everything I could about MPS but I also wanted the information to be current and up to date with the latest treatments. Since I could not find everything I wanted I decided to keep a diary of Chiara's journey through life and it became her online blog. Never did I imagine how many people would follow her life.

Last year I decided to enter a team into the Glasgow 26 mile Kiltwalk. This team was made up of family and friends all of who followed Chiara's life. Chiara's Clan was made up of 15 members including myself, her mum. January 2015 we all began our training. We knew that walking 26 miles would be hard but could it ever compare to what Chiara had been through in her 4 short years. Chiara had undergone 2 BMTs (bone marrow transplant), spinal fusion, adenoidectomy and tonsillectomy, was born deaf but now had normal hearing levels and on 11th March 2015 went through bilateral hip reconstruction and was in spica cast and was wheelchair dependent at the time of the Kiltwalk. With all these procedures in our heads we would wear our Kilts proudly and walk 26 miles.

The day was wet and dull but a great day was had by all. By lunchtime the sun was splitting the sky and all our spirits were high. As we approached the finish line we could see Chiara cheering us on and clapping. It made walking 26 miles all worth while. She crossed the finish line with each group of walkers in our team. Our last check of the fundraising page for the Kiltwalk was nearly €4000. Local shops and pubs also had collecting tins on display as Chiara had become a well known figure of Grangemouth. She is the lively, cheeky, fiery wee girl that everyone loves and copes fantastically well given everything she has come through.

I hope to complete the Glasgow Kiltwalk again next year but my ideal finish is for Chiara to walk across the line with me and no spica cast next year.

Michelle Petersen

A huge thanks to Chiara's Clan for their amazing fundraising!

You can read more about The Kiltwalk by visiting www.thekiltwalk.co.uk

Hannah's Quiz Night

In 2014 Hannah McGhin held an incredibly successful quiz night (featured in our Summer 2014 magazine) and she decided to do the same again this year. Hannah obviously has some great fundraising skills as she made an amazing £700 from the night! Well done Hannah and thank you to everyone for making the quiz such a success.

Hannah emailed in tell us a bit about the event:

"I got in touch with local businesses and friends and family in order to get prizes for the raffle and tombola, everyone was more than generous! The whole event was a great success all thanks to the kindness and generosity of my friends and family. They all keep telling me I should turn it into a yearly event as it was so successful"

Photo above: Hannah (left), with friend Heather Scott who suffers from MLIII

Marina & Friends

Superstar fundraiser Marina, along with her team of volunteers, are still going strong in their second-hand shop dedicated to raising money for MPSIII research. To date the little shop, located in Bristol, has raised a staggering £141,828-48 through the hard work and dedication of Marina and her team.

Marina recently wrote in to tell us that she recently received a kind donation of £20 from Saula Vodonaivalu Jr.Fifita on the occasion of his fifth birthday. He was adamant that Marina and the charity should share in his good fortune.

Saula Vodonaivalu Jr. Fifita is from Tonga and lives with his family in Bristol. The entire family have been extremely supportive of Marina, with mum, Elsa, providing time and working in the shop.

Thank you to Saula for his kind donation, and a big thank you to Marina for her amazing support.

Daniel's Charity Fete

On 17th May, Sandra and Rashpal Singh held their annual fete in memory of their son Daniel, who tragically passed away from MPSII Hunter disease. The proceeds were kindly donated to ourselves, Stars Playgroup and Baginton Fields School, both of which Daniel attended.

Sandra and Rashpal wrote:

"The day went really well; there was loads of activities, footie, face-painting, music and dance, stalls, all the things Daniel loved. The weather stayed dry, although a little cold, and Sarah from the Willink came down with her lovely daughter, Ellie, to run the hook-a-duck stall and the MPS stall. We are so grateful to them for taking the time to come all that way.

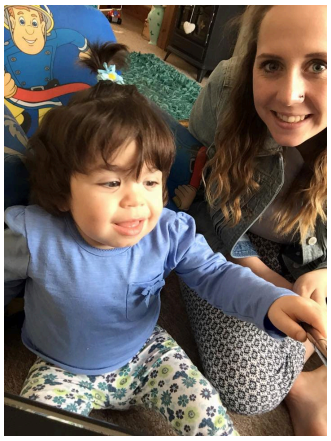
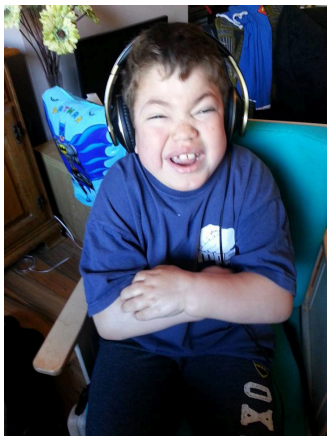
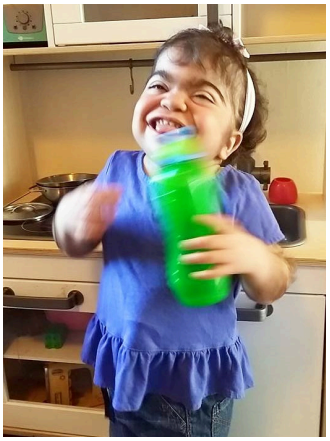
Everyone had a great time, lots of fun and laughter, just the right thing to remember our beautiful boy.

This was the third 'Daniel Charity Fete' so here's looking forward to next year."

Thank you so much to Sandra and Rashpal for donating £1537.50, which was raised from the fete. We really appreciate all your hard work and amazing support. Thank you also to Sarah and Ellie for manning the MPS stall!

Photos above show some of the brilliant activities on show at the fete.

Saula & family

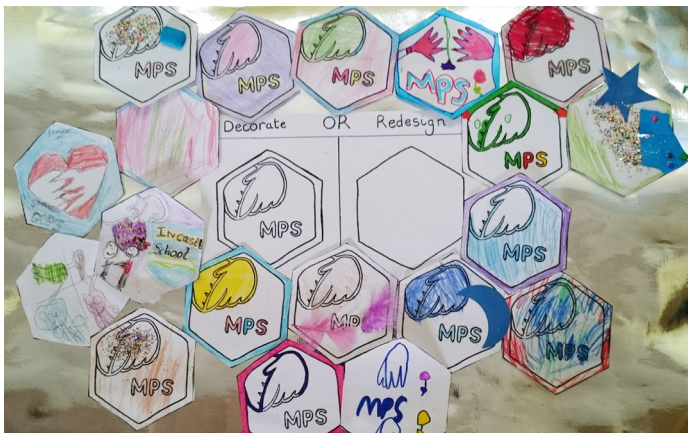





THANK YOU FOR WEARING IT BLUE FOR INTERNATIONAL MPS AWARENESS DAY 2015!

On behalf of the MPS Society, we would like to thank everyone who got involved with MPS Awareness Day 2015 on 15th May. From year to year we are astounded at how everyone comes together to raise funds and awareness on this special day, whether it is by Wearing It Blue, holding a blue cake sale, or any one of the other inventive ways you decided to mark the day. 2015 was no exception, and we were delighted to see so much activity throughout the day on our social media sites and email, with our supporters pulling out all the stops to make the day special - it was fantastic to hear from you all. Thank you all for helping to make rare diseases, like MPS, stand out.

Here are just some of your stories from the big day...

Castle Special School



The corridors were a sea of blue at Castle School on the 15th of May, all of our students proudly showing off their blue garments. Everyone was looking forward to the day ahead and the whole school was buzzing with excitement.

In the morning Primary watched a video in assembly, then we took a picture of lots of people wearing their bluest finery. During morning playtime Primary had a bake-sale; we think the cakes were everyone's favourite.

Secondary had a Decorate or Redesign the MPS Logo competition, everyone tried really hard and turned out some beautiful designs! Then Girly Club ran a bake sale for Secondary during their lunch time break and sold almost all of the cakes and biscuits that were left over. In the afternoon, Secondary had a special assembly and talked about the MPS Society and why they were all wearing blue.

We all had a fantastic day and are very proud to say that we raised £124.50 for the MPS Society. We look forward to celebrating Wear It Blue Day again next year!

Castle Special School, Cambridge

LG Optical

Following on from their festive raffle in aid of the MPS Society last year, precision optical engineers, LG Optical, have once again pulled out all the stops to support our cause by Wearing It Blue for MPS Awareness Day. The team raised £57, plus some text donations on the day.

As you can see from the photo, they really did take 'Wear It Blue' to a whole new level!

Thank you to LG Optical for their wonderful fundraising and for being such good sports!

Boudoir Wine Bar

Boudoir Wine Bar in Glasgow did a fantastic job of raising awareness and funds over MPS Awareness Day. As you can see, the French and Italian inspired bar produced some brilliant blue cakes and drinks, and made great use of our merchandise.

Owners of Boudoir, Darren and Amanda Scott, and parents to Sophia (MPSIII) also gained the support of Sophia's nursery (Accorn Park) and St Meddans Toddler Group. Altogether they raised a fantastic £1166.60, with further donations pouring in from friends and family.

Thank you to everyone who contributed to Darren and Amanda's fundraising!

Wear It Blue in Malawi

Last year we saw Raihana Seedat support Wear It Blue day at Growing Star Nursery School in Malawi, to raise awareness of MPS. This year she once again did a fantastic job, and emailed in to tell us how she got on:

MPS Awareness day at my son's school St Andrews International Primary school went very well.

As I got out of the car and walked out of the car park... it was surely a sight to see! Everyone dressed in blue...students teachers and parents. Some of the teachers were pointing out their blues to make sure I had noticed them. It was amazing. We surely raised awareness in Malawi, and we raised a total of £212.50.

Thank you Raihana and all who got involved in Malawai!

Ian Evans & Arriva The Shires

Long-term supporter and father to Harry (MPSI), Ian Evans once again got behind our Wear It Blue campaign, along with the rest of the staff at Arriva the Shires. Drivers were allowed to take MPS Society collection boxes into their buses on their route and Wore It Blue for the day to help raise funds and awareness around the local area.

Ian, himself, donned a wonderful Paddington bear outfit, which we think is his best costume yet!

Once again, a huge thank you to Ian, Ken Hargreaves and all at Arriva the Shires for their continued support and hard work on MPS Awareness Day. Altogether they made a fantastic £1072.61.

Teresa & Corey's Wear It Blue

Last year Teresa Jeffery, along with her son Corey (MPSI), held a spectacular coffee morning for MPS Awareness Day. This year Teresa and Corey outdid themselves with a plethora of fundraising activites, and generally did a fantastic job of raising awareness. Teresa emailed in to tell us more:

At Corey's school they had a wear it blue day on Friday 15th. They raised £243.15.

His friends were very proud to be wearing blue for Corey!

Corey's friend Amelia had her first birthday in April and instead of presents she asked for donations to The MPS Society. Corey loves giving Amelia cuddles.

Her mum gave us £40 for The MPS Society and a card which resonates with all of us - "Keep fighting"

Us mums dressed in blue too!

Blue cupcakes, courtesy of Karen's Cakes were sold at The Guardhouse Cafe on Berry Head in Brixham for the whole weekend and raised £84.60

On Saturday May 16th Corey's Mum - Teresa and Dad - Shane and friends Kate, Scott, Phil, Dave and Mark took to the quarry at Berry Head in Brixham to abseil 80ft. Overwhelmed by people sponsoring us there was most definitely no backing out! 80ft doesn't seem that high until you're at the top with just a rope between you and the bottom! Grenville House in Brixham provided us with an instructor and all the equipment. It was scary but great fun and we each had a couple of goes, the worst bit was leaning back at the top!

The one person I kept thinking of on the way down was Corey and that me being scared for a few minutes is nothing compared to what he has to go through on a regular basis. He is our inspiration.

Thank you to Teresa and her abseiling team, Karen's Cakes, Corey's school, Amelia, and of course Corey!

To date, Teresa has raised £1826.97 from her action-paked fundraising.

Shire Pharmaceuticals

Our thanks to the team at Shire Pharmaceuticals UK, who marked International MPS Awareness Day 2015 by wearing MPS Society t-shirts and collecting donations..

Barclays, Canary Wharf

Led by Relationship Director, Ian Baldwin, the staff at the Canary Wharf branch of Barclays Bank Wore It Blue, with the main star of the show clearly being Ian himself (pictured left), who had donned a very fetching ensemble of a blue wig, blue onesie and cowboy boots! We like!

A huge thank you to Ian and his team who raised a fantastic €321.17 for the MPS Society.

Penparc Primary School


Tuesday, May 26, 2015

Seeing blue as pupils raise cash to help combat ra

Pupils at Penparc Primary school Wore It Blue in support of Alicia Evans who attends the school.

Alicia suffers with MPSI Hurler and her kind-hearted fellow pupils wanted to help raise funds and awareness of her condition. The children did brilliantly and raised €245 and got a mention in their local press.

Alicia's mum recently made a video about Alicia's journey for MPS Awareness Day, which you can view at this address: https://flipagram.com/f/VZkXOuirTZ

eBay for Charity

As you will have been aware, we were kindly supported by eBay for Charity, who featured us at the checkout during the week of our MPS Awareness Day. Not only did this enable us to raise lots of awareness among the general public, but eBay shoppers also donated just over €5,476 to us over the course of the week!

Thank you to eBay shoppers for their generosity, and to eBay for Charity for giving us this brilliant opportunity!

Crockerne School Wear It Blue for Freddie

Freddie De Gennaro lives in North Somerset with his mum and dad and brother Joseph. He is 6 years old and has MLIII, Autistic Spectrum Disorder and epilepsy.

On Friday 15th May, all of the children at Freddie's school, Crockerne Church of England Primary, wore it blue to help raise money and awareness for Freddie's condition and other children like him.

Freddie Wearing It Blue!

The playground was a sea of blue faces, blue hair, blue ribbons - one of the Year 2 children said, "Even the sky is blue for Freddie's Day!"

A special assembly was held to talk about Freddie's condition and why the children were wearing blue, and they all applauded Freddie.

It was a great day and we were very touched by the generosity and the thoughtfulness of all the children and staff at the school. A massive thank you!

Justine De Gennaro
Freddie's mum

Thank you to Justine, Freddie and all at Crockerne School for their wonderful support and for raising a brilliant £300!

MPS Awareness Day at James Walker UK Ltd

Emily in her blue regalia!

Gayle, mum to Emily Bradshaw (MPSI), emailed in to tell us how her workplace, James Walker UK Ltd, helped to mark MPS Awareness Day. Thank you to all at James Walker for your support and to Gayle for doing an excellent job of raising awareness at work.

On the 22nd May here at James Walker UK Ltd we organised a Dress Down day and cake Sale for MPS Awareness Day. As we couldn't arrange this for MPS Awareness day on the 15th May I made sure everyone here had a little knowledge of the charity and conditions they support by posting some information on our company intranet.

I was overwhelmed by the generosity of everyone who contributed to the Dress Down day and cake sale. In total we raised €111.61. Everyone here at James Walker were so supportive and positive in their comments towards Emily and her progress made. I have only worked at James Walker for 7 months but the support has been outstanding.

Gayle Bradshaw

Wear It Blue for Ethan

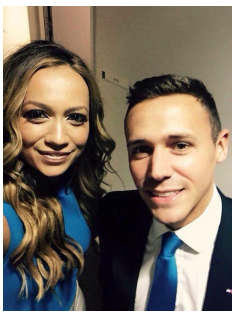
Ethan

Katrina Fanneran, mum to Ethan (MPSVI) and Trustee of the MPS Society, has once again pulled out all the stops for MPS Awareness Day. Friends, family, associates and even celebrities have all got involved in her Wear Blue for Ethan campaign, which proved to be hugely successful.

On her JustGiving page Katrina writes about the challenges and ordeals faced by Ethan:

"In all of this Ethan stands defiant. He is unaware of his condition but endures weekly and sometimes daily therapies like they are a piece of cake. He gets upset and frightened of course but a little encouragement from Mummy and lots of hugs and kisses and he soon comes round. He is the bravest little boy and he inspires me every moment of every day. He gives me something to live and fight for and he is my little soldier."

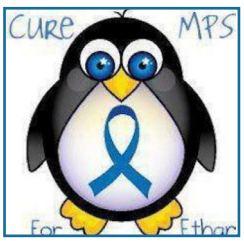
Stacey Solomon



Kate Abdo & Tom White from Sky Sports News Wearing Blue

For this year's MPS Awareness Day, Katrina raised a fantastic €5,640 on her JustGiving page - well done and thank you, Katrina!

https://www.justgiving.com/ethansmps/



Challenge for Fabry

On 23rd -24th May, my colleague Wil and I took on the London to Brighton Challenge; a 100km (62 mile) walk from Richmond to Brighton racecourse. My dad, sister, nephew and I all have Fabry Disease, so I had no hesitation in choosing the MPS Society as the charity I would fundraise for.

It was a clear sunny morning when Wil and I left Richmond with the other participants, perfect conditions. We enjoyed a scenic walk along the Thames for a while, and were in good spirits when we reached the first rest point. Two little girls clutched home-made 'Good Luck' signs and waved at us from their garden wall. Others along the way had set up tables with biscuits and squash! Participants were chatting to each other and a couple asked me what the MPS Society was, a chance to raise awareness! As the day went on, the terrain got rougher and aches and pains began to creep in. 7 hours and as many blister plasters later we finally got out of the M25 boundary. The route was very well signposted and we forgot our aching muscles momentarily as we saw the 50km marker. Half way!

6 km later as night was falling we reached a major restpoint where we were greeted by a jazz band and lots of hot food. The queue at the medical tent was long, and we saw that many people had to drop out, but we were lucky. All the training had paid off. Headtorches on, glowsticks attached and off we went into the night. The novelty of wearing headtorches soon wore off and the walk became more of a trudge. The dawn chorus we'd been so anticipating was just noise; we were that grumpy. Each kilometre marker seemed to be further and further apart. The real low point for me was the 80km rest stop. Everything hurt. I wasn't cold, but I was shivering and my stomach was churning. I overheard a medic (the support team was superb) tell another walker with similar symptoms to drink some squash. I took this advice and felt myself recovering. I remembered why I was doing this walk, had a pep talk with Wil and off we went again. Sometime later I slipped climbing over a stile. I wasn't hurt, but it took Wil and another walker to lift me, my legs were refusing to co-operate. Never mind, the end was near and we pressed on.

The final 10km was very picturesque. Our muscles were aching but I knew it was just a matter of putting one foot in front of the other and repeating. Wil's parents and sister were there at one point, cheering everyone on, it was heartening to see familiar faces. Up and over the Downs, and eventually Brighton Racecourse was in sight! At the beginning of the walk we'd jokingly discussed how we might sprint across the finish line, striking a pose; in reality it was a limp and a pained smile! Wil was stoic, but I really had to fight back a tear as we saw his family, my friend and complete strangers clapping and cheering for us. A glass of champagne and a medal later, it was over. 27 hours and 21 minutes from start to end. Easily the toughest physical challenge I have faced but so worth it. The cameraderie between participants was amazing, and hearing their reasons for walking was inspiring. I constantly reminded myself that I am fortunate to be well enough to attempt such a challenge and to have great friends and family to offer sponsorship and support! Almost £800 raised and still a few more donations to collect. At the time I said never again, but who knows...

Claire Harris

Clock Change Challenge for Jack

Back in March of this year, Nicky, along with her fiancé and friend, ran a 10k in memory of their friend Jack Stuart, who sadly passed away in December 2014 (see page 16 of our Spring 2015 magazine). Nicky emailed in to tell us about Jack and how her day went.

Jack was an inspiration, a lil person with the biggest heart and a bigger passion for life, to know him was to love him and love him we did, like so many!!! Losing Jack left a huge hole in the hearts of all who were blessed enough to have known him but also a hole straight through the heart of our community. Many of Jacks friends have gathered together to raise money in Jack's memory since his passing.

Back in January when we were all struggling to come to terms with the huge loss, I wanted something to occupy me through this hard time and The Clock Change Challenge 10k popped up. I have never been a runner and I thought this would be the perfect goal, which would also hopefully raise a good amount of money for the MPS Society, in Jack's memory. The MPS Society was so close to Jack's heart and has been so important to his family. I shared my plans with my fiancé, as Jack was his first friend since moving to Calne and meant so very much to him, so he decided to join me and at the same time Danielle decided to join us. Having three of us training made it alot easier! After contacting Jack's parents and checking they were happy for us to do it, we spoke with the MPS Society who were really supportive and sent us a sponsorship pack.

Luckily on the day all the training paid off and all three of us crossed the line together in 1 hour 9 mins, To be greeted by the very welcome sight of one of my children waiting for us with Manda and Emma Stuart, Jack's mum and sister, our very dear friends.

I cannot thank all our family and friends, Jack's family and friends and the MPS family enough for all your support and donations which enabled us to raise £770.32.

Nicky Mazzone

Great Manchester Run 2015

On 10th May 2015, a group of five MPS Society supporters completed the Great Manchester Run. Described as being Europe's biggest 10k running event, with over a quarter of a million people taking to Manchester's streets, taking in some of the city's most iconic buildings and locations, including the Old Trafford stadium and the Coronation Street set.

Thank you so much to our supporters for all your hard work training and fundraising for this event - you all did brilliantly!

Liz Stillo (pictured right) not only ran the 10k, but also held a fundraiser in her little village in the Peak District, which involved a homemade blue cake sale and a 'guess my time' competition.and raised almost £200, as well as a lot of awareness.

Thank you, Josie!

Josie Devlin, mum to Niall and Dermott, who both suffer with Morquio disease, recently retired from her job as Postmistress in Sheskinshule, Northern Ireland. Josie had run the local post office for 26 years, and in an incredibly generous gesture Josie donated £1,200 to the MPS Society.

Thank you Josie and we hope that you enjoy your retirement!

Dance Extravanganza for MPS

Over the weekend of the 16th and 17th of May, the Craddock-Hayes School of Dance hosted their annual dance show in Sittingbourne, Kent. This year's performance was welcomingly titled 'Be Our Guest'; a dance extravaganza with children aged between 4 and 23 dancing to the likes of Frozen, Ed Sheeran, Ellie Goulding and a number of musical classics in a combination of ballet, tap and jazz routines. This year also added the new dimension of adult dancers to the show's line up; ten lovely ladies performed a challenging and crowd pleasing tap number, much to the delight of the audience! A true variety showcase that perfectly combined talent with entertainment, the annual show weekend is the perfect opportunity for parents to see just how hard their children work.

Performed over two nights, 'Be Our Guest' was the culmination of several months work and a lot of dedication from pupils and parents alike. From attending weekly lessons to making costumes and practising choreography, there is a real sense of dedication within the Craddock-Hayes School of Dance. The show weekend always generates a real sense of camaraderie between all the pupils - young and old - and although the weekend is jam-packed, it's also the highlight of the year! Since establishing the dance school back in 1996, the Craddock-Hayes School of Dance has performed a grand total of 19 fantastic shows and every year seeks to donate all the profits from the show to a different charity. It has become a tradition the dance school is incredibly proud of and makes all the hard work leading up to the performances even more worthwhile.

This year CHSD managed to raise over £400 for the MPS Society, which is a charity especially close to one pupil's heart. Her family continue to have great support from the MPS Society so the Craddock-Hayes School of Dance took great pride in this selection. Despite the fact the school is growing in number year on year, there is a true familial feeling among its many members. Sue Craddock-Hayes (the Principal of the dance school) felt, as is often the case, that the donation feels all the more heart-warming when there are ties so close to home.

Sue was extremely proud of all her pupils and parents for all their hard work in the lead up to the performances, and was happy to see 'Be Our Guest' be such a success on stage. While pupils are currently working towards Medal Tests, there's already talk about next year's show which is likely to be an extravaganza as the school will be celebrating a massive 20 years of performances! With another year of hard work just around the corner, CHSD hopes that this year's efforts and all the money raised can be put to brilliant use at the MPS Society. On behalf of all the pupils and Sue herself, we wish you all the best.

Words by Jade Joseph, pupil since 1996

A huge thank you to all at the Craddock-Hayes School of Dance for their amazing fundraising!

London Marathon 2015

A huge thank you to all our runners, who trained, fundraised and finally ran the London Marathon for the MPS Society back in April. To date our charity runners raised an astounding £12,274 on their fundraising pages, with more to come, and we are so appreciative of their incredible support. Thank you Nicki, Lee, Nick, Kate, Jack, Carlos and Ben - you are amazing!

Featured below is Nicki Hare's account of the Marathon, which she ran along with her friend, Lee:

Team Effort to Cross the Line

I was lucky enough to gain one of the few charity places made available to the MPS Society for the London Marathon this year. It was my second marathon after completing London last year and I knew two things. Firstly if I was going to survive the winter training I needed the support of some great running and cycling friends to keep me company and motivated. Secondly I needed to run the race with someone. Running it on your own is lonely, despite the huge crowds so when my friend Lee told me she had secured a ballot place and that she would run for the MPS Society too we were away!

Training started in earnest just before Christmas, four runs a week were planned around work, family life and everything else that comes when you are the mum of four gorgeous children, two of which, Sophie & Tom, have Sanfilippo. January was a challenging month, chicken pox hit the house, Sophie had respiratory issues, Tom had surgery for an abscess in his mouth and I had a chest infection. The training plan took a bit of a hit but with only one full week missed there was more than enough time ahead to get ready. Each week the long runs got longer until they hit 20 miles and the interval training (to help with the speed) became less like intervals and more like a mental challenge of endurance! When we could, Lee and I ran together which was more often that we thought would be possible. Lee travels A LOT with work, Sau Paulo, Tokyo, San Francisco, Hong Kong, Dusseldorf, Oslo were just some of the countries she travelled to between January and March and I seriously don't know how she managed to fit in the training with the jet lag. Our running pace was so similar and we were excited to be running the marathon together, that was until we received our running packs in early March and discovered that Lee's number meant she would be starting in the blue start and mine the red one. What difference does that make? Due to the sheer volume of runners, they follow a completely different course for the first three miles so the chances of us meeting one another and running together (hand in hand across the line) faded overnight. It was a major emotional setback for us both but we kept up the training and convinced ourselves that running on our own would be just fine!

The big day came and in the weeks leading up to it we were both overwhelmed by the kindness of friends, family and beyond who had so generously donated to our Just Giving page. We had raised nearly €5,000 between us so not finishing the race, despite Lee carrying an injury was not an option!

Proudly wearing our MPS Society vests and matching shorts we headed off to Greenwich and said our goodbyes on Blackheath. 'See you at the end ' I called as I wandered off in the rain to the mass red start. The atmosphere was amazing, despite the cold wind and rain and I spent the next hour hearing stories from others and sharing mine.

At 10.15 on 26th April the gun went and we were off. I tackled many demons over the next 3 hours and 38 minutes. This year I loved the crowd, I made friends along the way, including superman who I ran with for some time, I enjoyed running through Canary Wharf (a low point the year before)....and we both did it, with only four minutes time between us! In fact our half marathon times were only 20 seconds apart so whilst we weren't physically together during the race, we were mentally there, along with Sophie and Tom who we carried on our shoulders and whose strength of dealing with Sanfilippo each day drove us across the finish line.

> *"This year I loved the crowd, I made friends along the way"*

There are so many people to thank. Both Paul's (husbands) for juggling the children across the winter, our wonderful children themselves for not moaning at our absence, our fantastic family and friends who cheered us on far and wide, on the day and the months leading up to it and lastly Rachel and Alison, who made sure Sophie and Tom were thoroughly loved and well looked after on the day. As for next year, we'll have to wait and see...

Nicki Hare

Thank you to all our donors including...

Children at **Naburn C.E. Primary School** wore blue in support of Blake's Genes and raised £45 for the MPS Society.

Claire and James Garthwaite sent in a further £155 towards their 'Not the Antiques Roadshow' fundraiser.

MPS Society Trustee, **Wilma Robins**, recently celebrated her birthday and asked for donations to be made in lieu of presents, collecting a total of £150. Trustee, Judith Evans donated a further £25 for Wilma's birthday.

Staff at **William Anelay**, building and restoration contractors, raised £131.78 by wearing blue for Blake's Genes day, which took place on Friday 13th February.

Gracie Mellalieu (MPSIV) and her fellow pupils at **Southdown Primary School** held a Wear It Blue fundraising day and raised an amazing £650!

Jacqui Mount and a group of friends, otherwise known as Team Jordan, are running the Glasgow 10k, and as part of the sponsorship raised Jacqui sent in £250, which was donated by a local businessman.

Dave Chapman at Arriva The Shires held a raffle and raised £110 for the MPS Society.

Ken Hargreaves conducted a staff survey at Arriva The Shires, earning £100, which he then donated.

Lee Skelton, Emma Bowers and Sara Staveley completed the Major Series North, a run and obstacle course. The team managed to raise a combined total of £651 in support of Blake's Genes.

Zoe Payne and Craig ran the Hastings Half Marathon and raised £132.50 on their Justgiving page.

Darryl Brook, Jack Severy and Mark Anderson ran the Brentwood Half Marathon. The team each set up JustGiving pages and managed to raise a combined total of £1750.10. The team fundraised in support of Darryl's son, Dylan, who was recently diagnosed with MPSIII.

Kerri Shaw ran the Hastings Half Marathon and raised £491.25 on her JustGiving page.

Sarah Pearson completed the Major Series North and raised £706.25 on her JustGiving page.

As featured in our Summer 2014 MPS Magazine, **Leon Batchelor** broke the pain barrier and completed the Virgin London Marathon 2014, raising an absolutely incredible £6130.75 on his Virgin Money Giving page.

Staff at the **Royal Bank of Scotland**'s Credit Documentation department nominated the MPS Society as one of the charities they are to support throughout the year. The team recently raised £586.21 from a raffle and collection.

Michelle Freeman and Sarah Williams held a vintage sale and tea party and raised £165. The pair wanted to support Isaac Turner (MPSI).

Staff at **Adrian Flux Insurance Services** held a dress-down day in aid of the MPS Society and raised £578.82.

Jackie Page held an art sale in support of Jack Watson and raised £338 for the Genistein Trial.

Holly Rowsell took part in a charity boxing match in support of Blake's Genes and raised £418.75 for the MPS Society.

Lymington Infants School held an 'Easter Community Tea', and sold some Easter gifts, with the proceeds amounting to a total of £142.50. The Easter Gifts, knitted Easter chicks filled with chocolate eggs, were hand-knitted by Nancy Lance.

Paul Knight ran the Hastings Half Marathon and raised £35 on his JustGiving page.

David Horner's grandson **Guy Horner**, and his partner Harriet Benison, raised £150 for the MPS Society by running the Manchester Marathon.

Mrs M Brock's kind friend **Barbara Marsh** has collected £7 by saving 5p pieces and copper coins for The MPS Society since Mrs Brock's granddaughter was diagnosed with MPS.

Adam Lloyd ran the Brighton Marathon and raised a total of £301.25. Adam is friends with a family who have been affected by MPSIII Sanfilippo.

Claire Hilton, a friend of Nicky Hare, completed a marketing survey on Facebook for a free donation of £100 to The MPS Society.

Charleswood Estates Ltd donated an amazing £5,000 toward Richard Vickery's London to Paris cycle.

The Lions Club of Paignton kindly sent in a cheque for £100, which had been raised by Tracey Wealthall who had been sponsored to take the challenge of walking into the sea on Boxing Day 2014. Brrrr!

Henson Hotel held a charity night and raised £310 for the MPS Society.

Father and Daughter **Mick and Kayla Yates** took part in the Grand East Anglia Run, raising a total of £400.71 for The MPS Society. Kayla's son, Mick's Grandson, suffers from MPS II Hunter's Syndrome.

Kate Stone raised £3700 for the MPS Society by running the 2015 London Marathon.

Nicholas Brooker ran the London Marathon raising £147.50 for The MPS Society. Nicholas' niece Lily suffers from MPSIII Sanfilippo.

Ben Whitnell went for a second medal attempt at the 2015 London Marathon, and managed to raise £551.38 for The MPS Society.

Jack Bovey ran the 2015 London Marathon and raised £1833.28 for The MPS Society.

Charlie and Sam Dodd raised £1356.39 for The MPS Society by completing the Lichfield Half Marathon.

Richard Newson very kindly donated £50 in support of a relative that was recently diagnosed with MPS III.

Rachel Boome donated £10 for sponsorship supporting Freddie De Genarro on a Wear it Blue Day.

Samantha Page donated £10 for sponsorship supporting Bobby Gill on a Wear it Blue Day.

Sarah Gill donated £5 in support of her nephew Bobby on a Wear it Blue Day.

The Rotary Club of Wallasey and an anonymous donator within the Club have funded a volunteer for a child from Wallasey to attend the 2015 MPS Conference at the cost of

Donations

The Eveson Charitable Trust; Mrs A. Baker; Angela Weedon; Victoria Fairweather; The Whartons Primary School; The Bown Family; Loraine Mimer; Sheanin Hughes; Elizabeth Eaton; The Shauna Gosling Trust; The JTH Charitable Trust; The Martin Connell Charitable Trust; Mrs A Baker; St. Mary's Gillingham Green Masonic Lodge; St. James the Less church wives group; Georgina Starling; Fay Bouri; Tony Moore; Mufaddal Najefy; Arlene Murray; Jackie Sumner; Raymond Webber; Rachel Todd; Susan Clarke; G Plummer; Graham & Margaret Moore; Lloyds Bank Foundation; Mrs Anne Cock; Mr M. Ismail; K. Carter; Bournemouth & Poole Medical Society; Savills PLC; Mrs Crotty; Elizabeth Gill; Richard Newson; Sarah Holland-Leppard; Rita Dulson; Samantha Lazenby; Paul Andrea; Swift Locksmiths; Mr T Lavelle; Mrs Linda Rowland; The Baily Thomas Charitable Fund; The Clover Trust; Blaenbaglan Primary School; Penparc Community Primary School; Jessica Reid; Kay Humphries; Donatas Mikuta; Mrs Elizabeth Herapath; Mrs J E Slocombe; Kate Shipman; Capital International Ltd; Sonya Munn

In Memory

Jack Stuart; Mrs Eileen Drayne; Mr Mark Cambridge; Christopher Croft; Mrs Mary Blackburn; Mrs Julie Hopper; Timothy Stansfield; Jess Robertson; Matthew Hardy; Tom Hibbert; Deirdre Coles

Collection boxes, stamps, foreign coins, mobile phones, ink cartridges, jewellery

Coffee Station; Sue Hollidge; Elegant Cuisine Ltd; Ian Evans and Arriva The Shires

The Society would like to thank the following donors for their regular contributions by either Standing Order or Give As You Earn

R & K Dunn; S Littledyke, Norman Saville; M Newell, Marcia Tosland, S Bhachu; C Cullen; S Brown; I & A Hedgecock; V Lucas; S Winzar; D Forbes; P & R Shrimpton; G Simpson; D & S Peach; C & M Gibbs; Mr & Mrs Cock; A Dickerson; Manjit Kalsi; D J Holmes; P J Martin; P Summerton; A Weston; E White; CL Hume; A Byrne; Mrs D M Robinson; S Cadman; A Sullivan; J & F York; J Wilson; J & M Wood; A Tresidder; K Robinson; K Osborne; Molly Rigby; Mr Thompson; E Cox; M Peach; C Garthwaite; Raymond Arnold; J Ellis; I & V Pearson; William Cavanagh; A Sabin; Barbara Harriss; L Brodie; A Ephraim; J Dalligan; M Malcolm; E Mee; Mr Hahner; K Brown; E Moody; E Brock; M Fullalove; G Ferrier; E Parkinson; Margaret Leask; R Taylor; R Gregory; L Stillwell; R & K Henshell; K & S Bown; S & J Home; V Little; M Reeves; S & D Greening; Z Gul; J Casey; J & V Hastings; J Winzar; Daniel Winzar; E M Lee; K Seeber; L Twaddle; Michael Morris; Abby Thomas; Elizabeth Merryweather; J Garthwaite; Nick Miles; Peter Rennoldson; Matt Mould; Michelle Boxell

£300! Their contribution will help to enable this young boy from Wallasey to enjoy a wonderful weekend of fun while his parents benefit from the information and social aspects of the conference programme.

Sarah Fournier raised £176.25 by walking the Just Walk 40km in The South Downs in support of 2 year old Dylan who suffers from MPS III Sanfilippo.

Eliza Turner and her friend Emma raised 788.75 in a 5k Junior Run in support of Eliza's brother Isaac who suffers from MPS I, Hurler disease.

Jessica Hooper and her colleagues raised £402.50 by running The Watford 10k in support of Jamie, who suffers from Sanfilippo disease.

Matt Boggan completed the 2015 Isle of Wight Challenge, raising an amazing £2176.55 in support of Blake's Genes.

Richard Vickery, Alan Dixon, Robert Finch and Michael Weeks completed the London to Paris Bike ride to raise a fantastic £15617.15!

The Rainbow School for Autistic Children raised £122.59 by holding a Wear it Blue day.

Deborah and Mark Burniston, Michael Riley and Michael Maule raised a fantastic £1537 by holding The Burniston Fun Run 2014 and completing last year's Great Manchester Run and The Great North Run.

Susan Joseph's Dance School put on a dance show to raise £405.

Luke Bown's School held a Wear it Blue Day on MPS Awareness Day and raised a brilliant £110.

Graham Martin raised £100 on a Wear it Blue day in support of a friend and colleague that suffers from MPS.

Morgan O'Hara raised £347.00 by running the Great Manchester Run for his nephew Sam that suffers from MPS.

Tracy Gibson raised £517.72 by running the Great Manchester Run for Sophie and Tom who both suffer from Sanfilippo (MPSIII)

Ashley Dodd, Daniel Williams and Keiran Andrews cycled up and down Snowdon's Ranger Path to raise £448.75 for Fabry Disease.

The Whitecliff Surgery in Dorset have raised £92.50 by holding a Wear it Blue Day on MPS Awareness Day.

The Brislington Art Group donated £30 to Marina and Friends following a recent fundraising event by The Art Group.

Josephine Murty, Sandra Irvine, Jacqueline Mount, Nicola Kelly, Fiona Holdsworth, Emma Morrice, Gillian Kelly and Kari Struthers Butler all ran The Glasgow 10k Run raising a fantastic grand total of £3276.80!

Marcia Burnett raised £382.60 from selling donated items and £77.50 by organising a sponsored walk in pyjamas!

Darryl Brook raised £92 by holding a Wear it Blue day at LV Ipswich.

Mrs Elizabeth Mee held a coffee morning and raffle in South Hiendley Parish Hall to raise a wonderful £260.41.

Katrina Gedge donated £410 raised from a Bingo evening with fish and chip supper and raffle.

Acorn Nursery raised £266.70 from a Wear it Blue Day. Sophia Scott attends the Nursery and suffers from Sanfilippo.

Meddans Baby & Toddler Group held a wear it blue day and raised £200.00 on behalf of Sophia Scott.

Christadelphian Church Sunday School collected £18.71 from the children that attend.

Mrs & Mrs McGrattan have kindly donated £350 to thank The MPS Society for all their support and advice given to their family over the years for their daughter Catherine.

Hayyan Hussain kindly donated £94.51 raised from a cake sale. Hayyan is 5 years old from Slough and suffers from Morquio disease.

Dorothy Robinson raised a wonderful £220 by holding a Card Workshop in support of Hannah Shannon who suffers from MPS III Sanfilippo.

Sarah Robson and the Team at RBS Bank raised £23.85 by wearing it blue in RBS Sunderland Branch. Sarah's school friend Gemma has a son named Jack who suffers from Sanfilippo.

The Rotary Club of Crewe & Nantwich Weaver very kindly donated £300 to The MPS Society for the cost of a conference volunteer for a child in their area.

Liz Still raised a total of £197.50 by running Manchester Great Run.

Elizabeth Gill donated £500 that was raised from the wedding of Laura Williams and Chris Grover in lieu of wedding favours. Congratulations Laura and Chris!

Gemma Corder ran the Bupa 10k with David and Michael and they raised £614.98.

Jessica Bumby cycled from Leeds to Liverpool, which is a total of £127 miles! Jessica raised £1233 from this challenge.

South and Lincoln teamed together to raise £112.50 which will be shared between The MPS Society and Frimley Park Hospital. They raised this money by completing The Junior Open Water Triathalon at Eaton Dornay and The Frimley Park Fun Run.

The Rotary Club of Mansfield donated £200 towards the cost of a volunteer for our 2015 MPS Conference.

The Reading Dispensary Trust donated £300 towards the cost of a volunteer for our 2015 MPS Conference.

Matt Fowler and his team completed a The Yorkshire Three Peaks Challenge to raise a wonderful £1606.25 in support of Blake's Genes.

Mark Butler and his team cycled a fantastic 86 miles on their New Forest Spring Sportive Epic Sunday even and raised a wonderful £250.

THESE BOOTS ARE MADE FOR FUNDRAISING

Wicked Walkabout

Have fun, get fit and help support those affected by MPS, Fabry and related diseases by holding a Wicked Walkabout this year. All you need to do is get together with some friends, family or work colleagues, decide the route for your walk and start getting some sponsorship money in!

The great thing about this event is that you can make it what you want, whether you fancy a midnight walk around your local town or a forest hike the choice is yours. You can even decide how you would like to fundraise - you could ask each person for an entrance fee or a minimum sponsorship.

Just remember to request your fundraising pack and let us know what you are planning by emailing fundraising@mpssociety.org.uk or give us a call on 0345 389 9901. MPS T-shirts are available from our online shop and we have lots of free fundraising materials to help you on your way.

Help us to continue supporting the children and adults affected by these devastating diseases and funding vital research into treatments by walking for MPS!

Dorset Wicked Walkabout

Once again the Wicked Walkabout Queen, Kath Hiller, has struck again! Kath held her third Walkabout for MPS on 17th May, accompanied by a group of friends and family, which took them from Bridport, along an old railway line to the beaches of West Bay (Broadchurch in the TV series) and back again.

Kath, who is grandmother to Joseph (MPSII), raised an incredible total of £872.96 with the help of her old school-friend, Pauline. A huge thank you to everyone involved and especially to Kath for such wonderful support of the charity.

Raising and Giving on Campus

Does your University hold a RAG (Raise and Give) Week?

Many universities have RAG committees dedicated to supporting local and national causes. Here at the MPS Society we want to raise awareness, fund vital research and continue to support our MPS Children and families. You could support us by:

- Holding a Rag Raid
- Organise a Ball or Club Night
- Sponsored World Record attempts
- Jailbreaks – get sponsorship to get as far away from your university and back in a set amount of time without spending any money
- Waxing, dyeing, shaving, painting yourself in the name of charity!

Sign up with us today and help support The MPS Society – and if there is no committee, then why not create one!

Let us know how we can support your application and together we can make a difference to MPS sufferers' lives.

For more information and fundraising materials please call Emma on 0345 389 9901 or email e.henry@mpssociety.org.uk or visit our website www.mpssociety.org.uk.

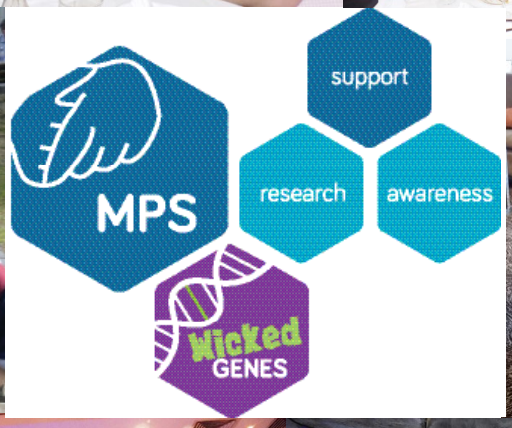








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